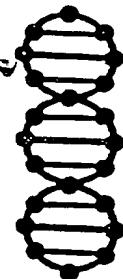
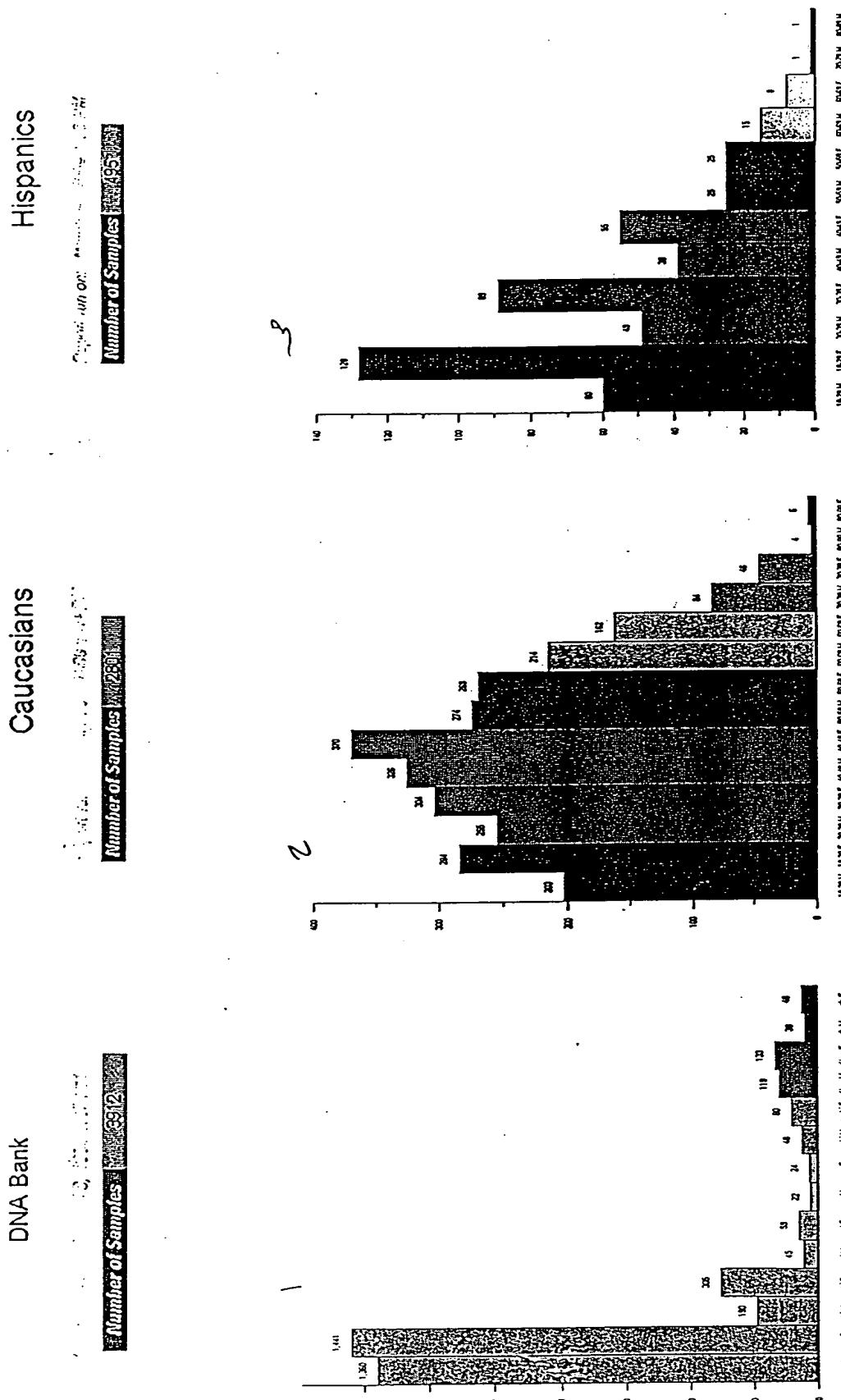


# Sample Bank

00001 001 00000 00000 00000



SEQUENOM  
INDUSTRIAL GENOMICS



1000 1001 1002 1003 1004 1005 1006 1007 1008 1009 1010 1011 1012 1013 1014 1015 1016 1017 1018 1019 1020 1021 1022 1023 1024 1025 1026 1027 1028 1029 1030 1031 1032 1033 1034 1035 1036 1037 1038 1039 1040 1041 1042 1043 1044 1045 1046 1047 1048 1049 1050 1051 1052 1053 1054 1055 1056 1057 1058 1059 1060 1061 1062 1063 1064 1065 1066 1067 1068 1069 1070 1071 1072 1073 1074 1075 1076 1077 1078 1079 1080 1081 1082 1083 1084 1085 1086 1087 1088 1089 1090 1091 1092 1093 1094 1095 1096 1097 1098 1099 10100 10101 10102 10103 10104 10105 10106 10107 10108 10109 10110 10111 10112 10113 10114 10115 10116 10117 10118 10119 10120 10121 10122 10123 10124 10125 10126 10127 10128 10129 10130 10131 10132 10133 10134 10135 10136 10137 10138 10139 10140 10141 10142 10143 10144 10145 10146 10147 10148 10149 10150 10151 10152 10153 10154 10155 10156 10157 10158 10159 10160 10161 10162 10163 10164 10165 10166 10167 10168 10169 101610 101611 101612 101613 101614 101615 101616 101617 101618 101619 101620 101621 101622 101623 101624 101625 101626 101627 101628 101629 101630 101631 101632 101633 101634 101635 101636 101637 101638 101639 101640 101641 101642 101643 101644 101645 101646 101647 101648 101649 101650 101651 101652 101653 101654 101655 101656 101657 101658 101659 101660 101661 101662 101663 101664 101665 101666 101667 101668 101669 1016610 1016611 1016612 1016613 1016614 1016615 1016616 1016617 1016618 1016619 1016620 1016621 1016622 1016623 1016624 1016625 1016626 1016627 1016628 1016629 10166200 10166201 10166202 10166203 10166204 10166205 10166206 10166207 10166208 10166209 10166210 10166211 10166212 10166213 10166214 10166215 10166216 10166217 10166218 10166219 101662100 101662101 101662102 101662103 101662104 101662105 101662106 101662107 101662108 101662109 101662110 101662111 101662112 101662113 101662114 101662115 101662116 101662117 101662118 101662119 1016621100 1016621101 1016621102 1016621103 1016621104 1016621105 1016621106 1016621107 1016621108 1016621109 1016621110 1016621111 1016621112 1016621113 1016621114 1016621115 1016621116 1016621117 1016621118 1016621119 10166211100 10166211101 10166211102 10166211103 10166211104 10166211105 10166211106 10166211107 10166211108 10166211109 10166211110 10166211111 10166211112 10166211113 10166211114 10166211115 10166211116 10166211117 10166211118 10166211119 101662111100 101662111101 101662111102 101662111103 101662111104 101662111105 101662111106 101662111107 101662111108 101662111109 101662111110 101662111111 101662111112 101662111113 101662111114 101662111115 101662111116 101662111117 101662111118 101662111119 1016621111100 1016621111101 1016621111102 1016621111103 1016621111104 1016621111105 1016621111106 1016621111107 1016621111108 1016621111109 1016621111110 1016621111111 1016621111112 1016621111113 1016621111114 1016621111115 1016621111116 1016621111117 1016621111118 1016621111119 10166211111100 10166211111101 10166211111102 10166211111103 10166211111104 10166211111105 10166211111106 10166211111107 10166211111108 10166211111109 10166211111110 10166211111111 10166211111112 10166211111113 10166211111114 10166211111115 10166211111116 10166211111117 10166211111118 10166211111119 101662111111100 101662111111101 101662111111102 101662111111103 101662111111104 101662111111105 101662111111106 101662111111107 101662111111108 101662111111109 101662111111110 101662111111111 101662111111112 101662111111113 101662111111114 101662111111115 101662111111116 101662111111117 101662111111118 101662111111119 1016621111111100 1016621111111101 1016621111111102 1016621111111103 1016621111111104 1016621111111105 1016621111111106 1016621111111107 1016621111111108 1016621111111109 1016621111111110 1016621111111111 1016621111111112 1016621111111113 1016621111111114 1016621111111115 1016621111111116 1016621111111117 1016621111111118 1016621111111119 10166211111111100 10166211111111101 10166211111111102 10166211111111103 10166211111111104 10166211111111105 10166211111111106 10166211111111107 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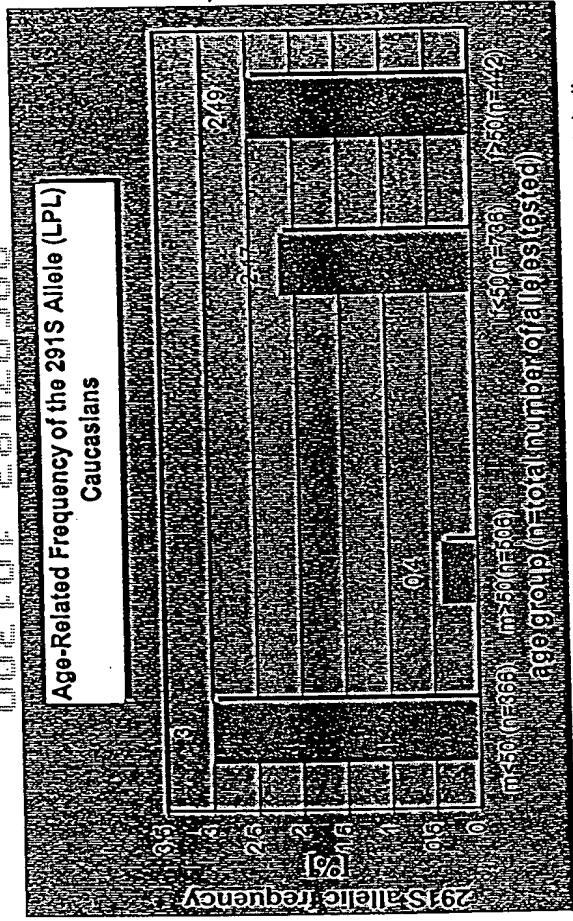


FIGURE 2A

age- and sex-distribution of the 291S allele of the lipoprotein lipase gene. A total of 436 males and 589 females were investigated.

Age-related distribution of the 291S allele of the lipoprotein lipase gene within the

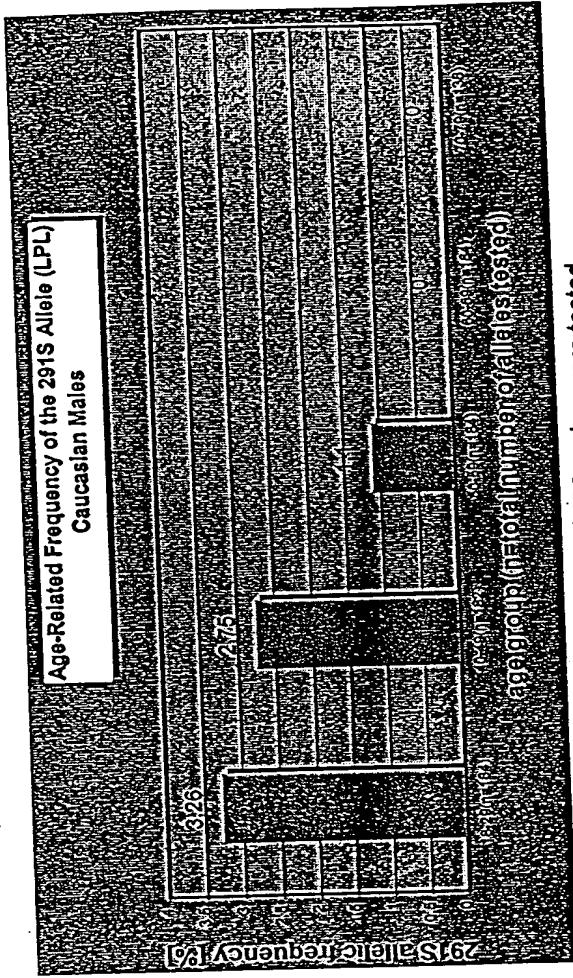
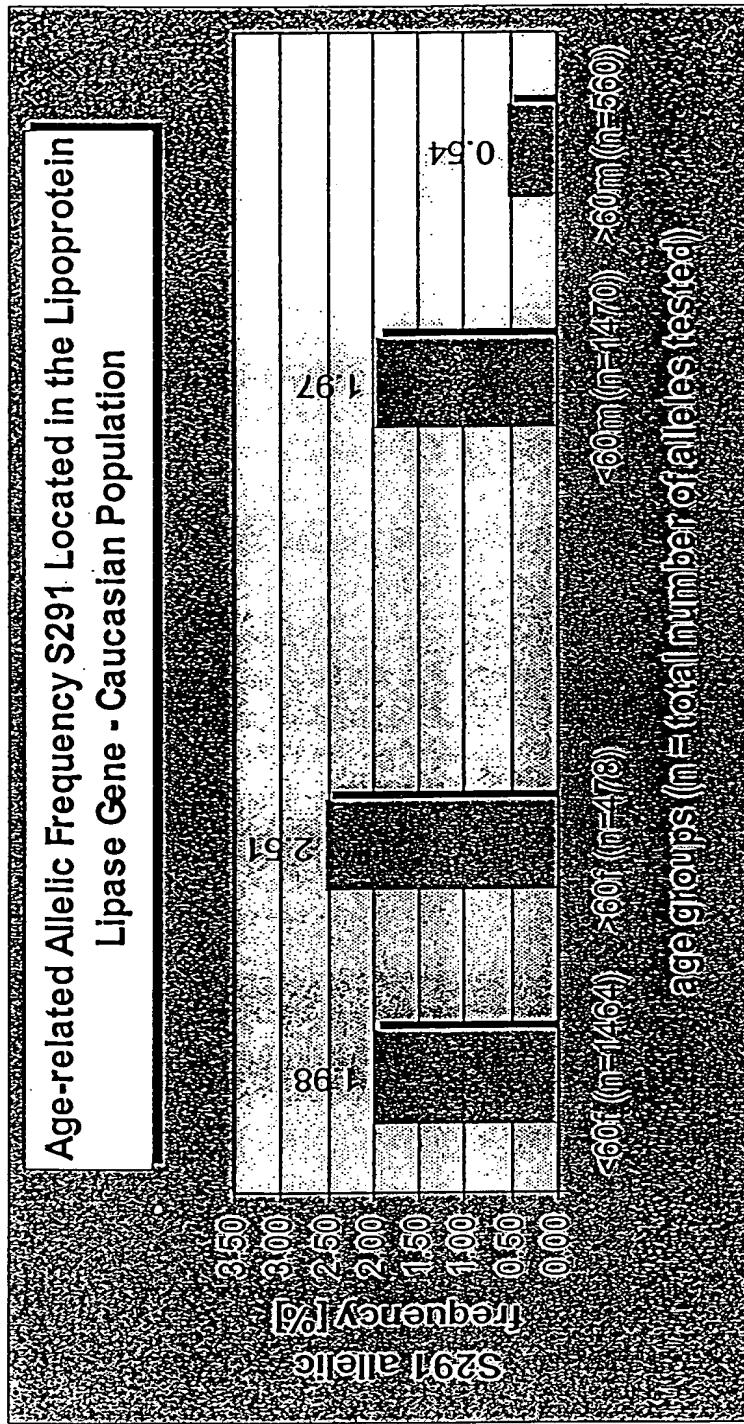


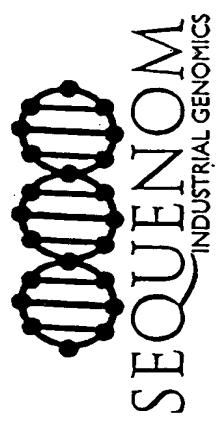
FIGURE 2B

male Caucasian population. A total of 436 males were tested.



Significance: Allele frequency of Ser291 drops from 1.97% to 0.54% in males;  $p=0.009$

FIGURE 2C

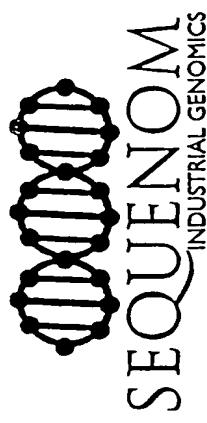


# Intelligent Genomics

# Questionnaire for Population-Based Sample Banking

### FIGURE 3

ONE FORTY-EIGHT MILLION



# Intelligent Genomics

## Sample Banks

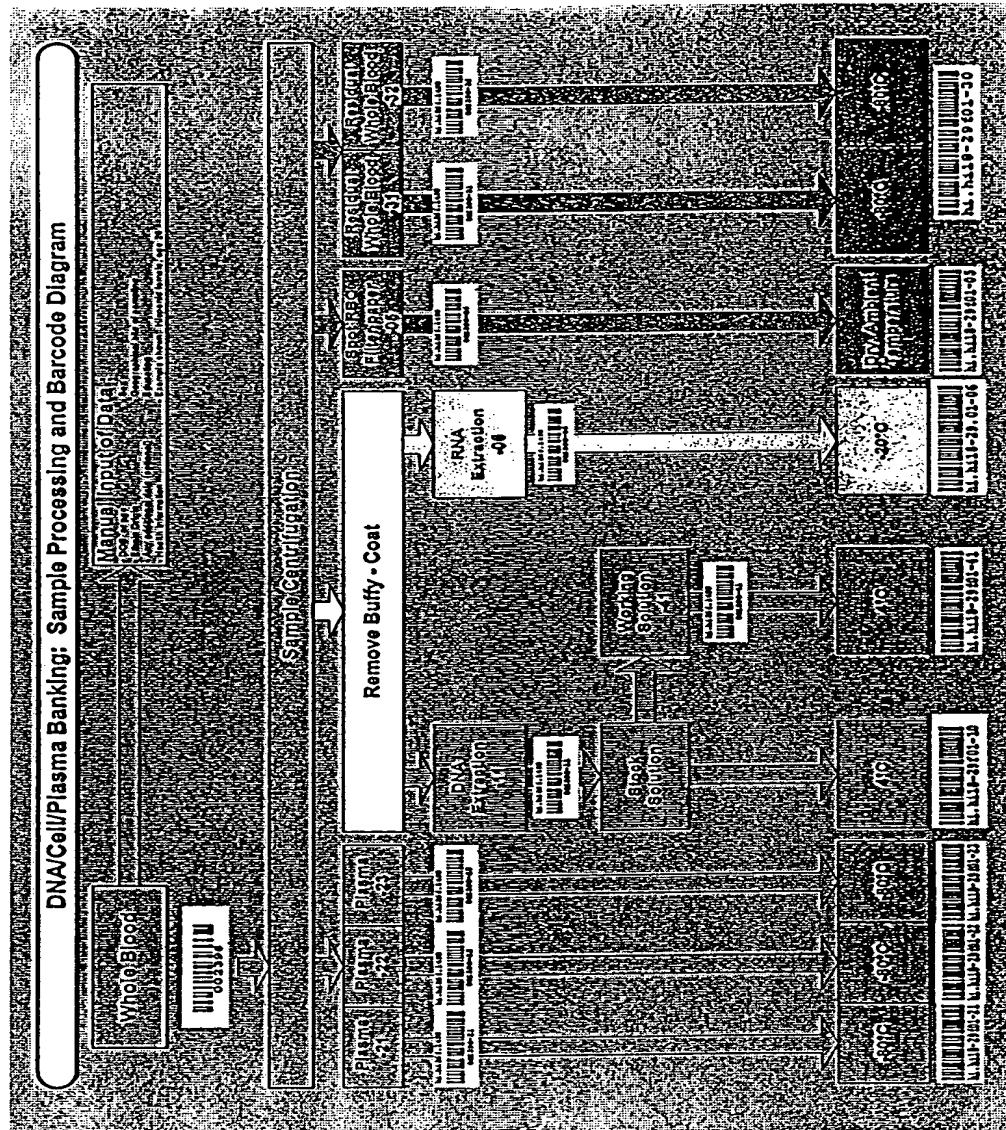


FIGURE 4

# Intelligent Genomics

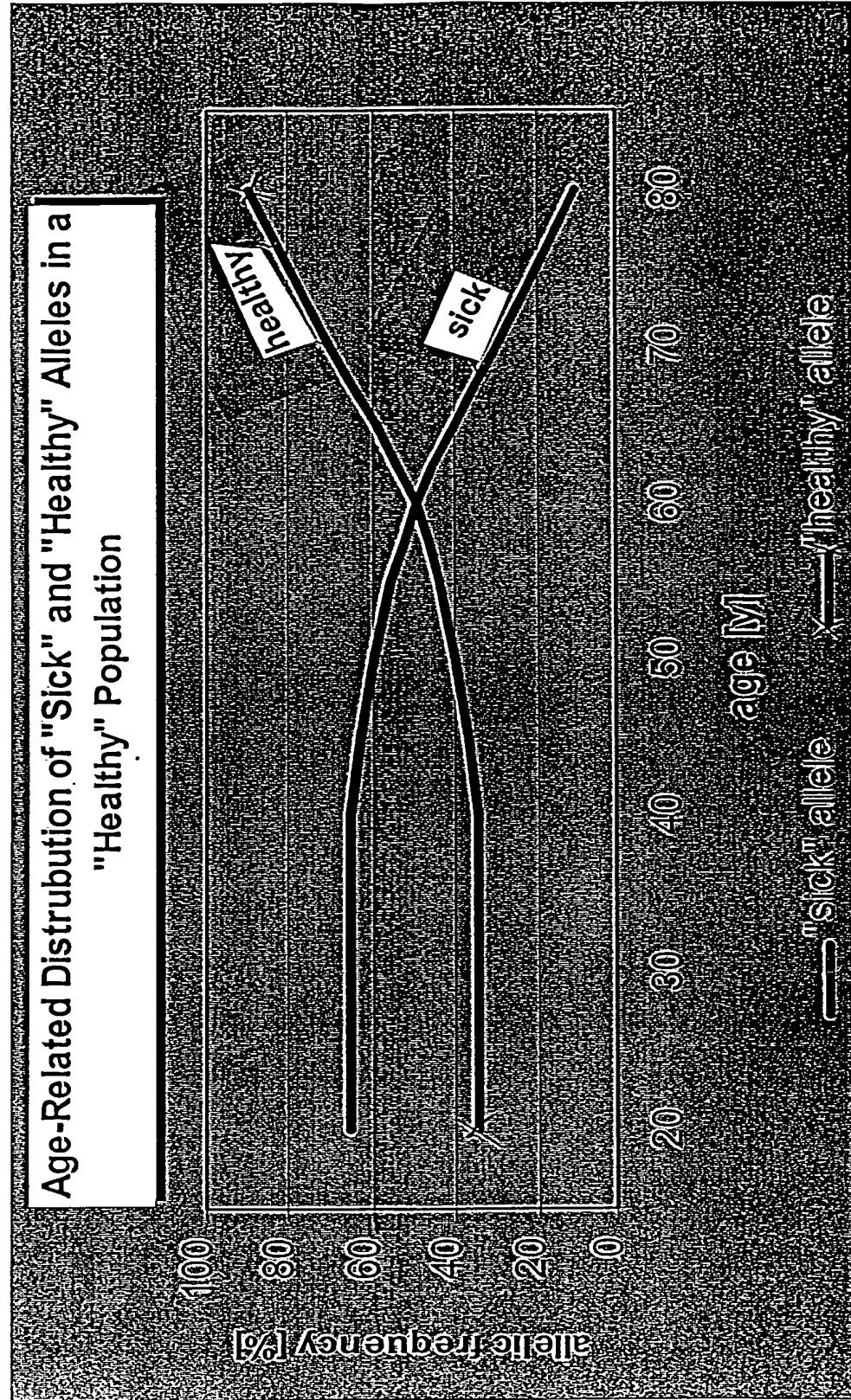
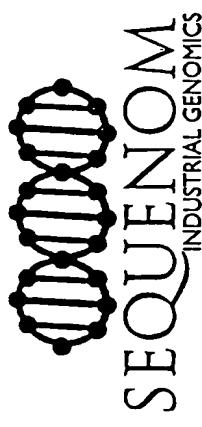
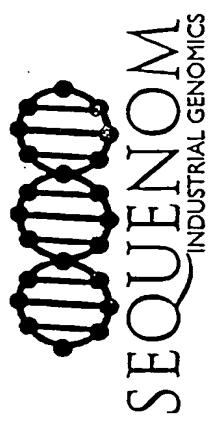


FIGURE 5

# Intelligent Genomics



## Age-Dependent Distribution of ApoE Genotypes / Alleles

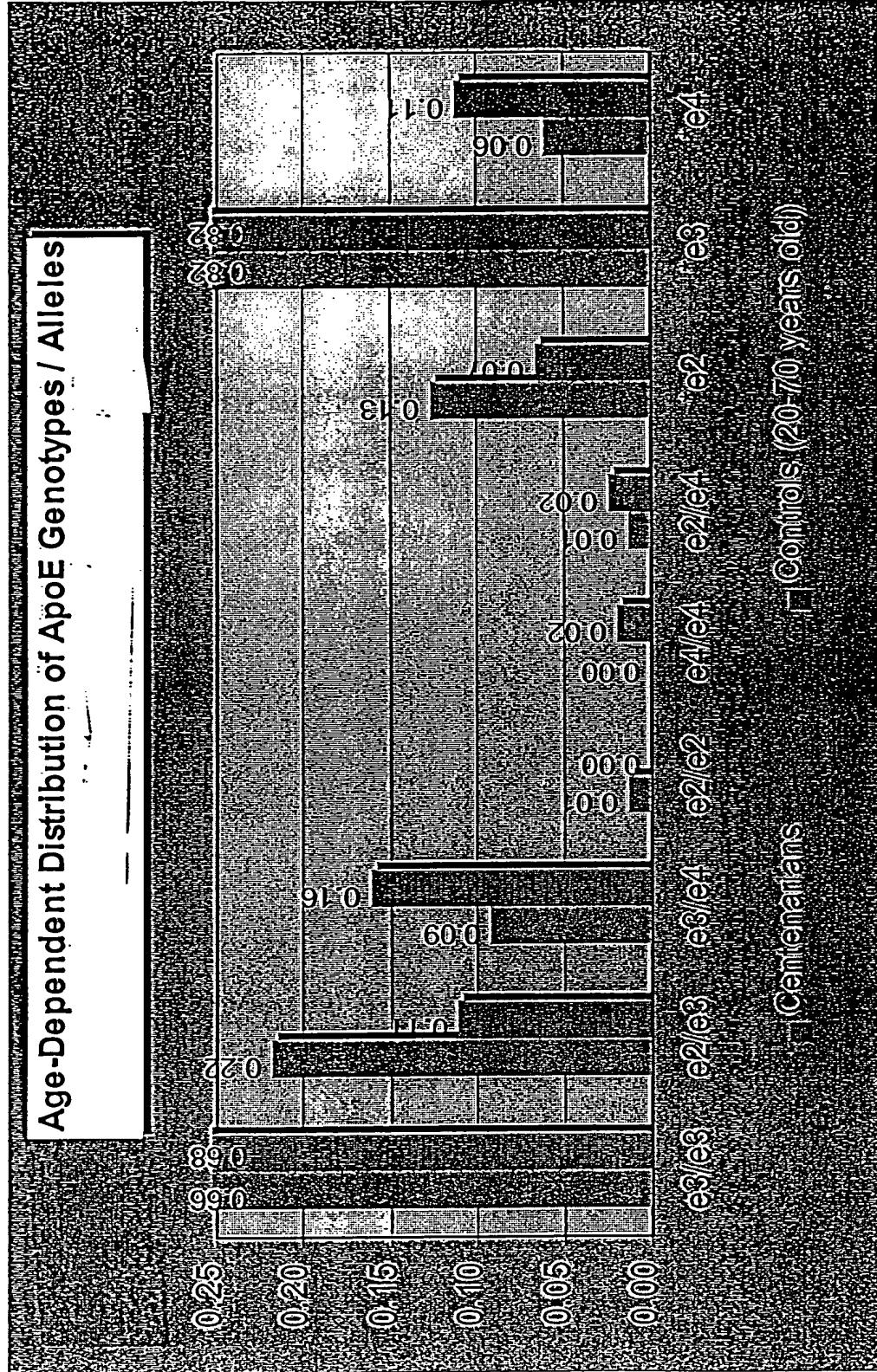
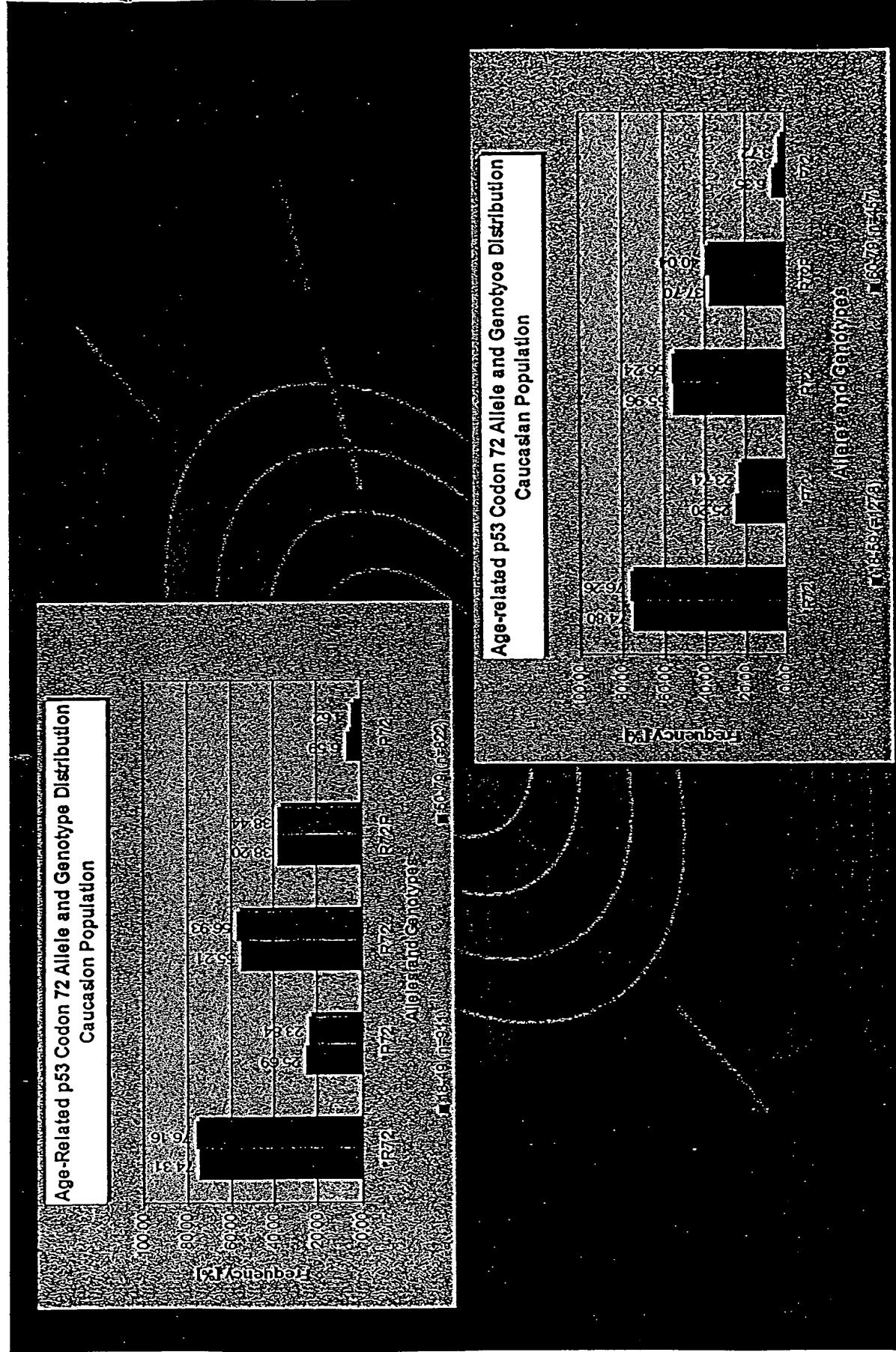


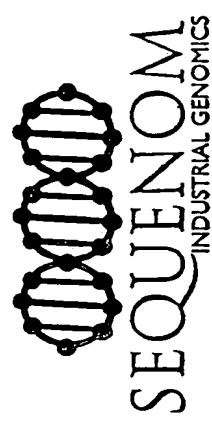
FIGURE 6

FIGURE 7A



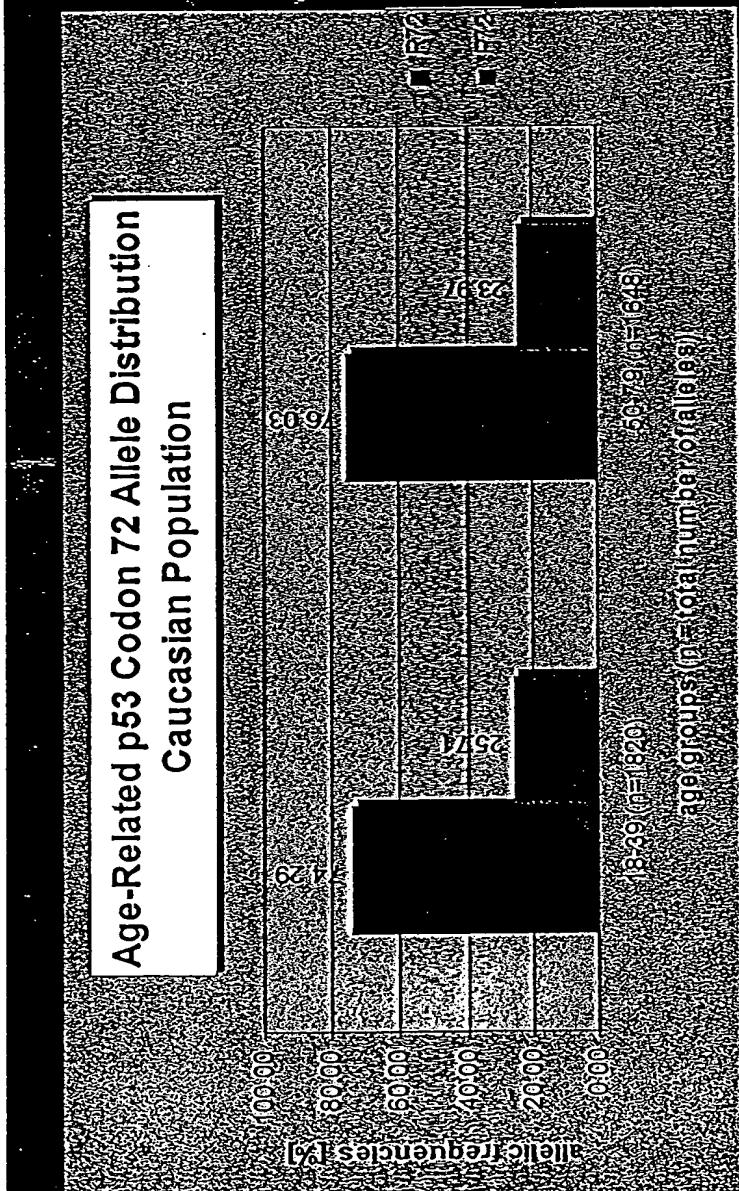
i Genotypes





CONTENTS OF THIS ISSUE

i genetics



**FIGURE 7B**

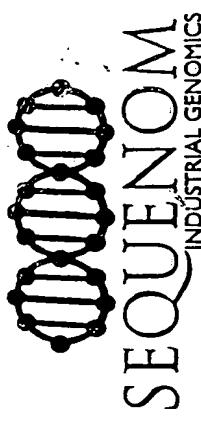


FIGURE 7C

i genetics

## P53 PP vs. PR/RR Genotype Distribution

By Age Cut point = 59

Genotype Freq (%)

PR/RR

N

Age Group

93.4

1278  
6.7

18-59

96.3

457  
3.7

60-79

Sample Size : 1735  
 $\chi^2 : 5.2$  (1 d.f.), P = 0.02

OBJECT "E84422950

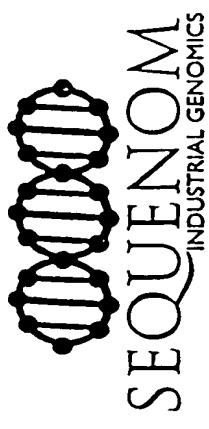
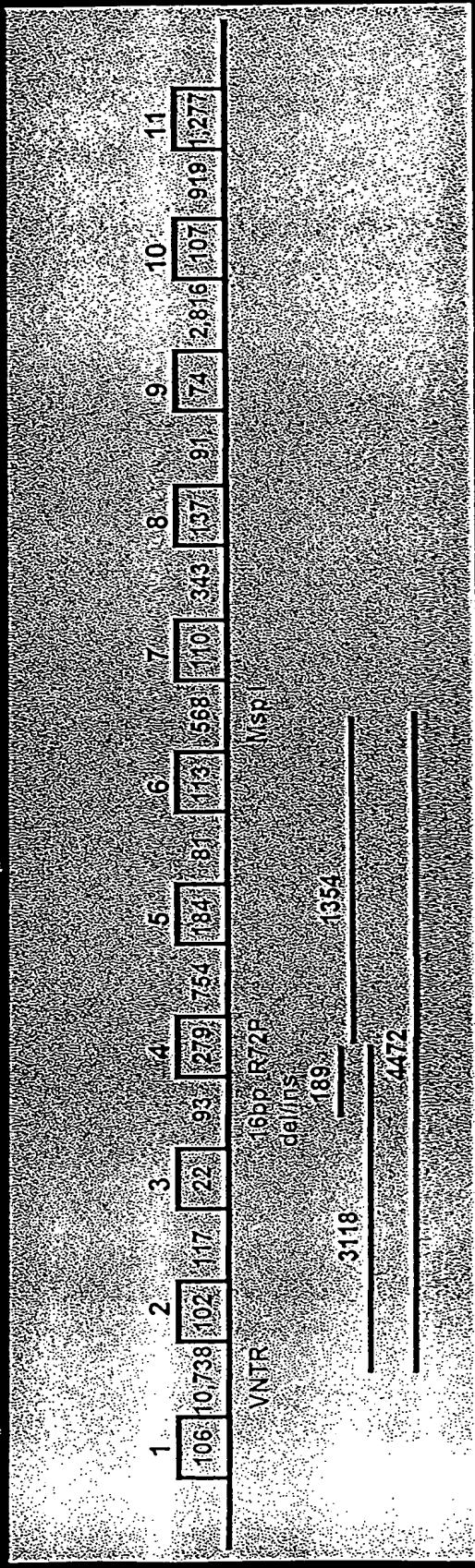
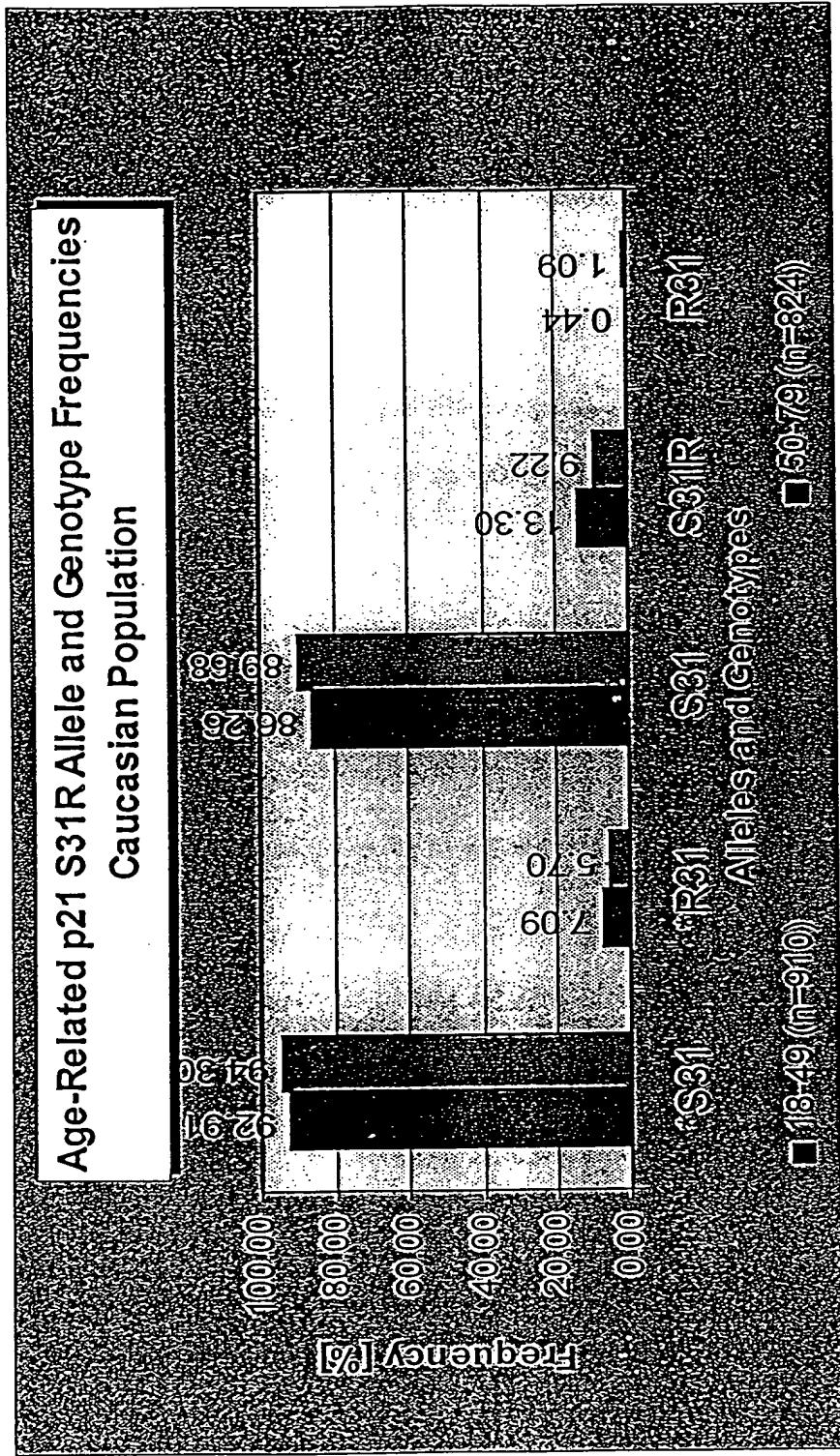


FIGURE 7D

## Genomic Organization of the p53 Gene



L. G. S. et al.



Significance: Genotype frequency of SR heterozygous drops from 13.3% to 9.2%;  $p=0.009$

FIGURE 8

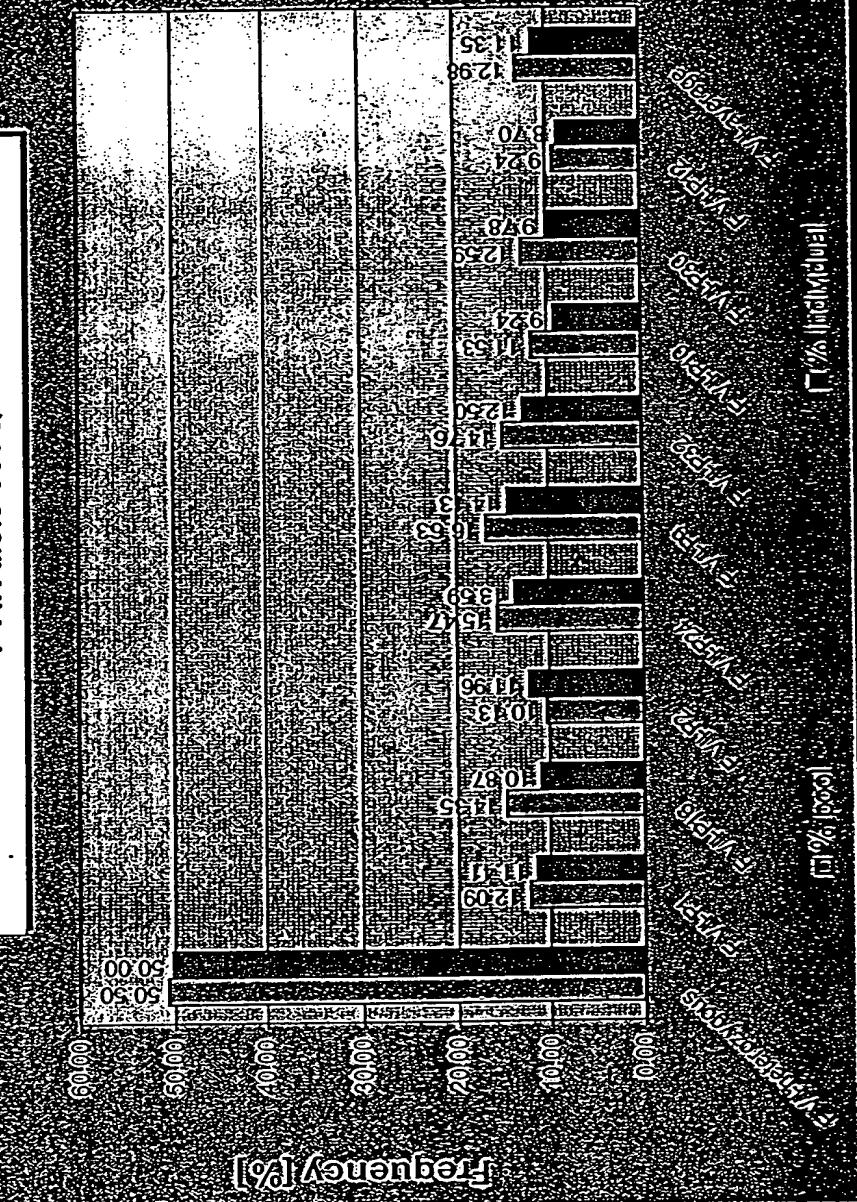
# DNA MassArray™

## FVII R353Q



SEQUENOM  
INDUSTRIAL GENOMICS

Allelic Frequency In Pooled vs Individual Samples  
FVII Allele 353Q



卷之三

# DNA MassArray™ CETP I405V

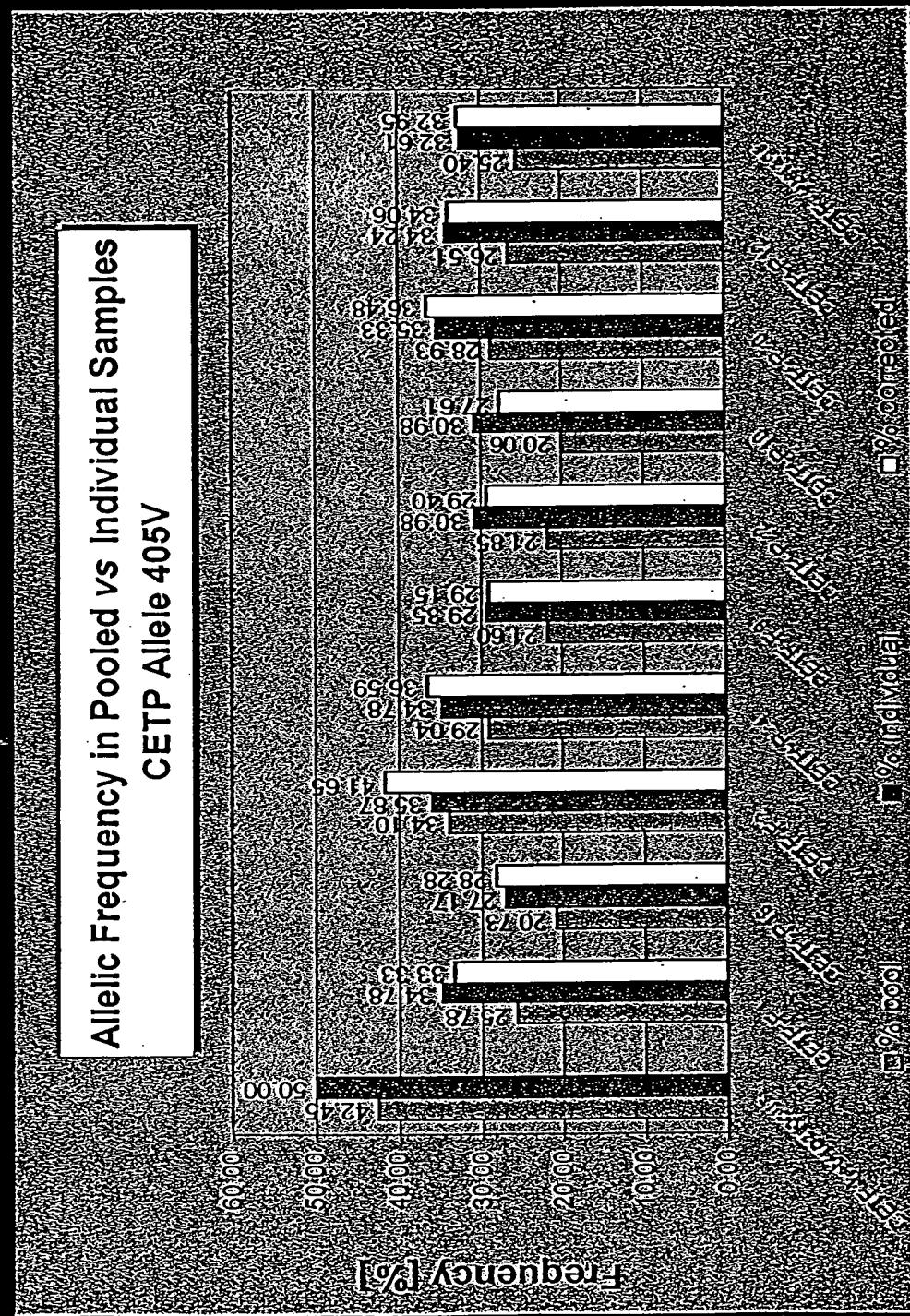
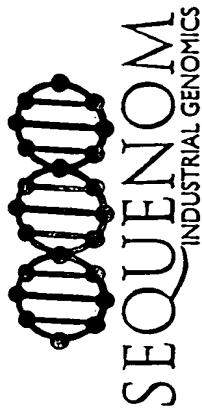


FIGURE 10

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DNA MassArray™

PAI-1 4G/5G

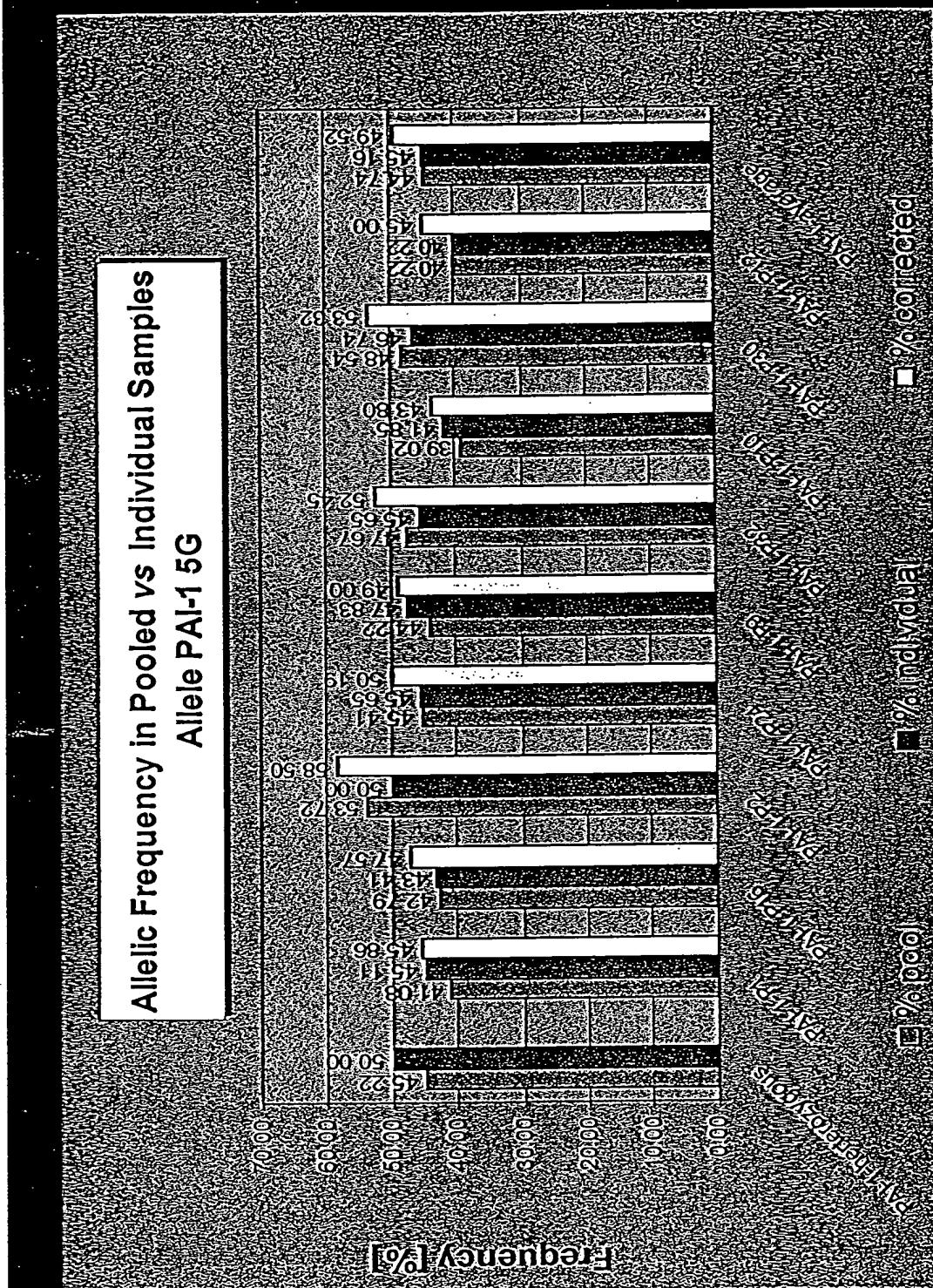
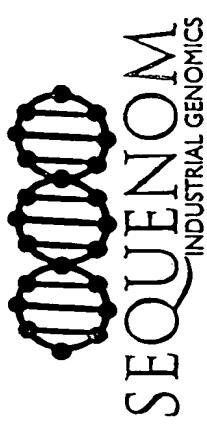


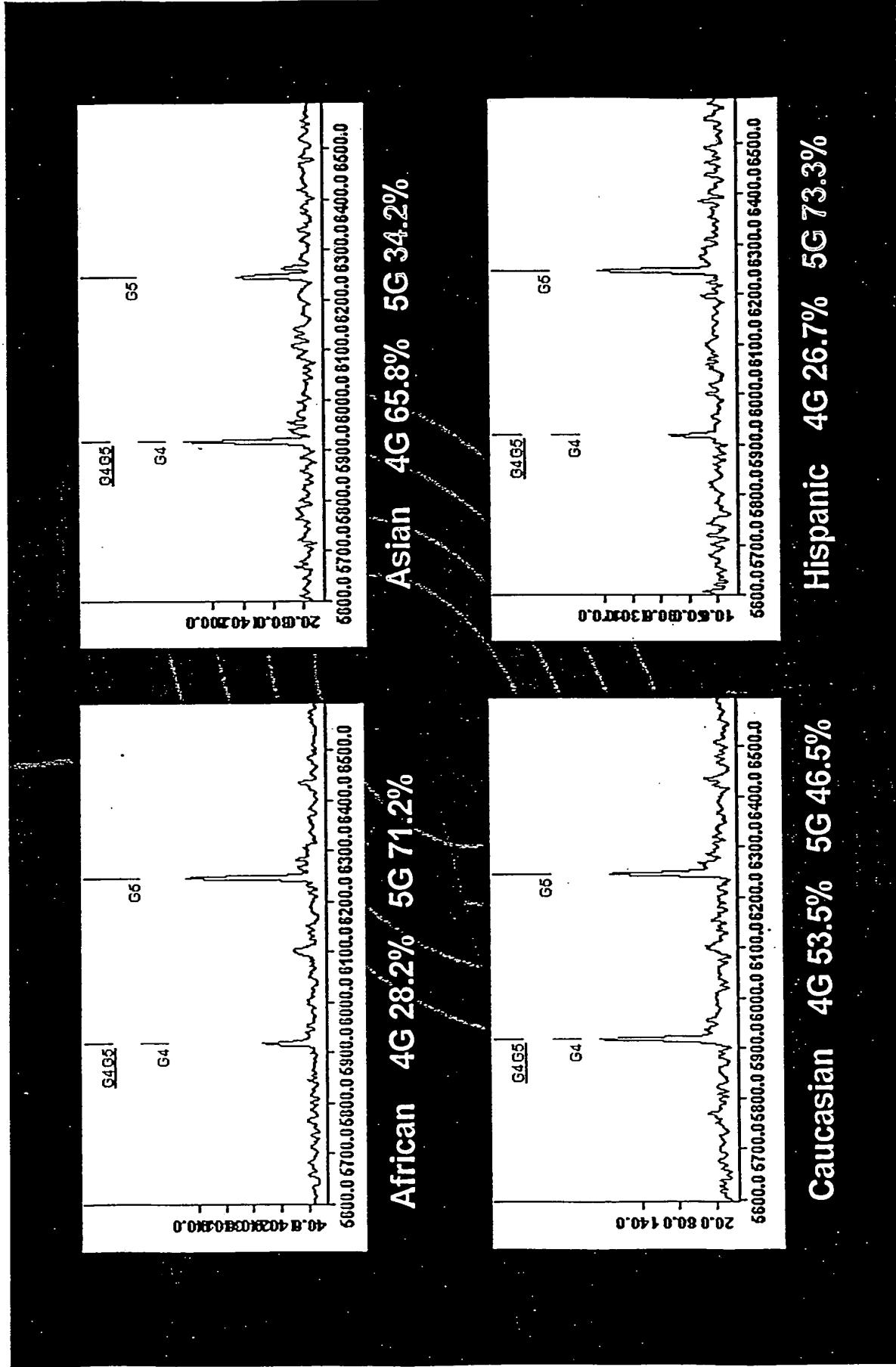
FIGURE 11

# DNA MasssArray™

## Ethnic Diversity (PAI-1)

 SEQUENOM  
INDUSTRIAL GENOMICS

FIGURE 12



# DNA MassArray™

## Ethnic Diversity (CETP 405)

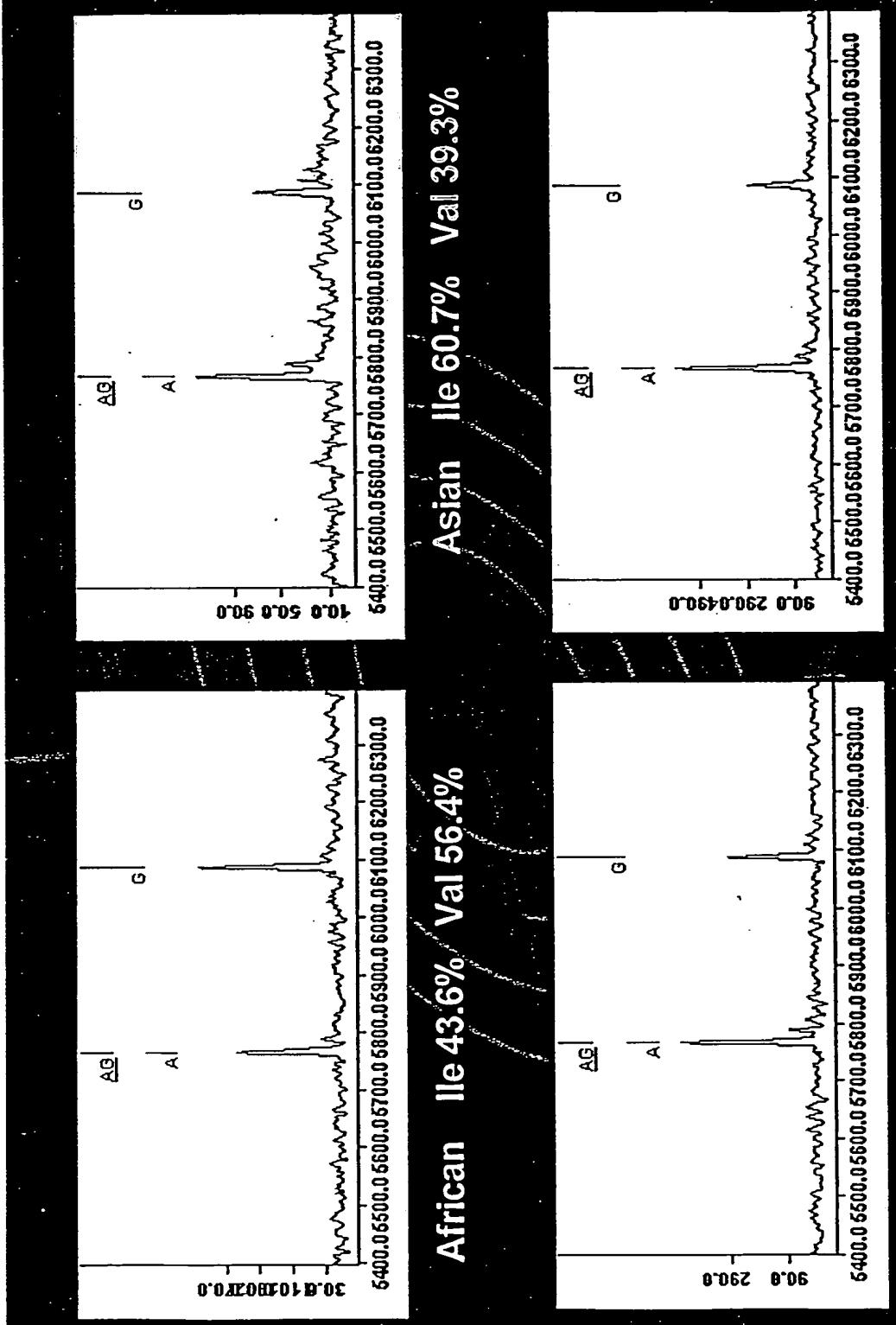
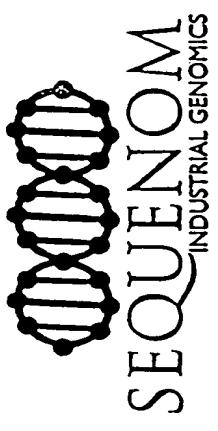


FIGURE 13

# DNA MassArray™

## Ethnic Diversity (Factor VII 353)

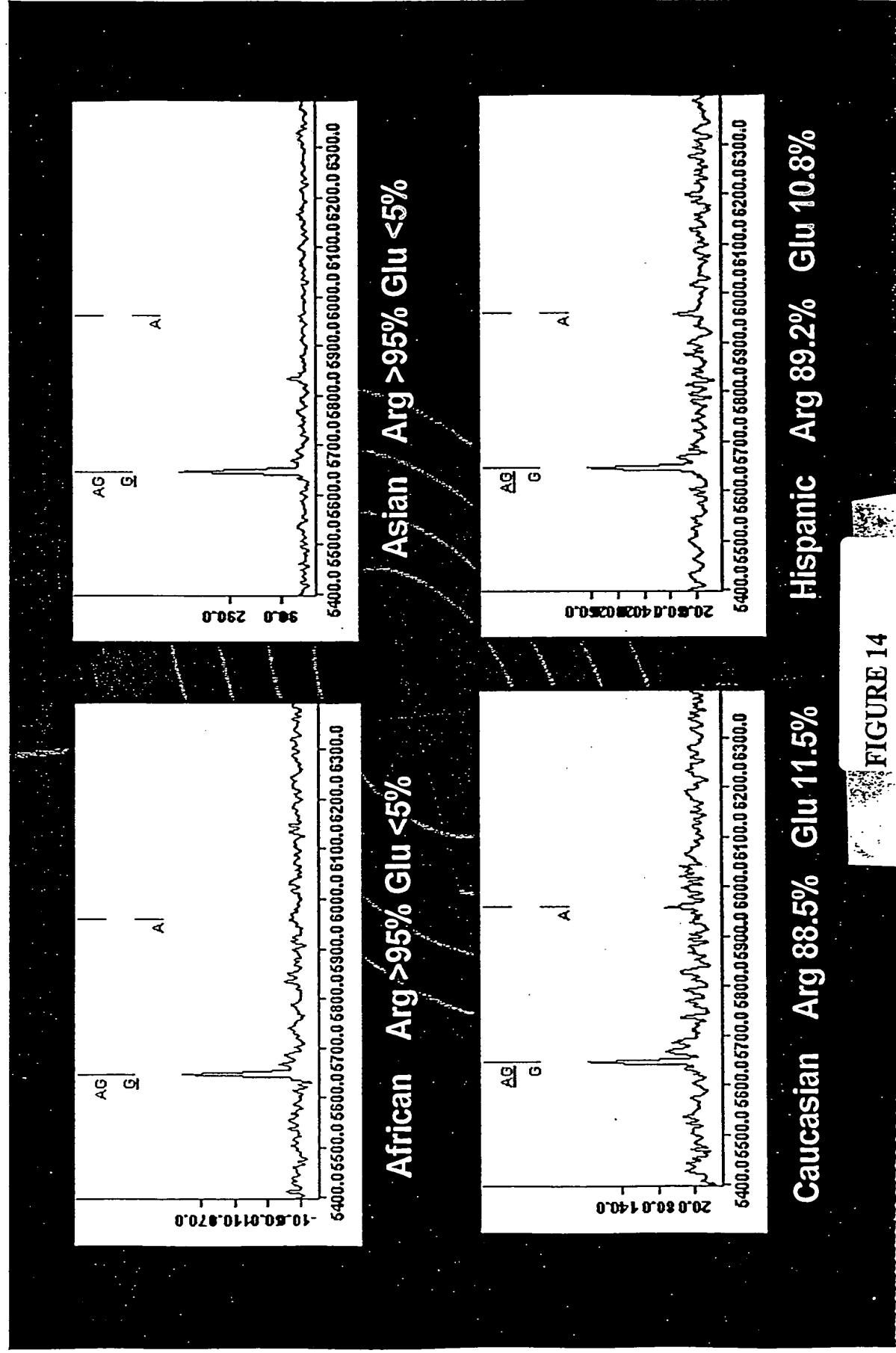
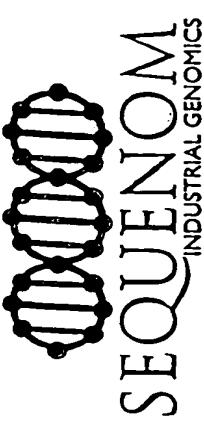
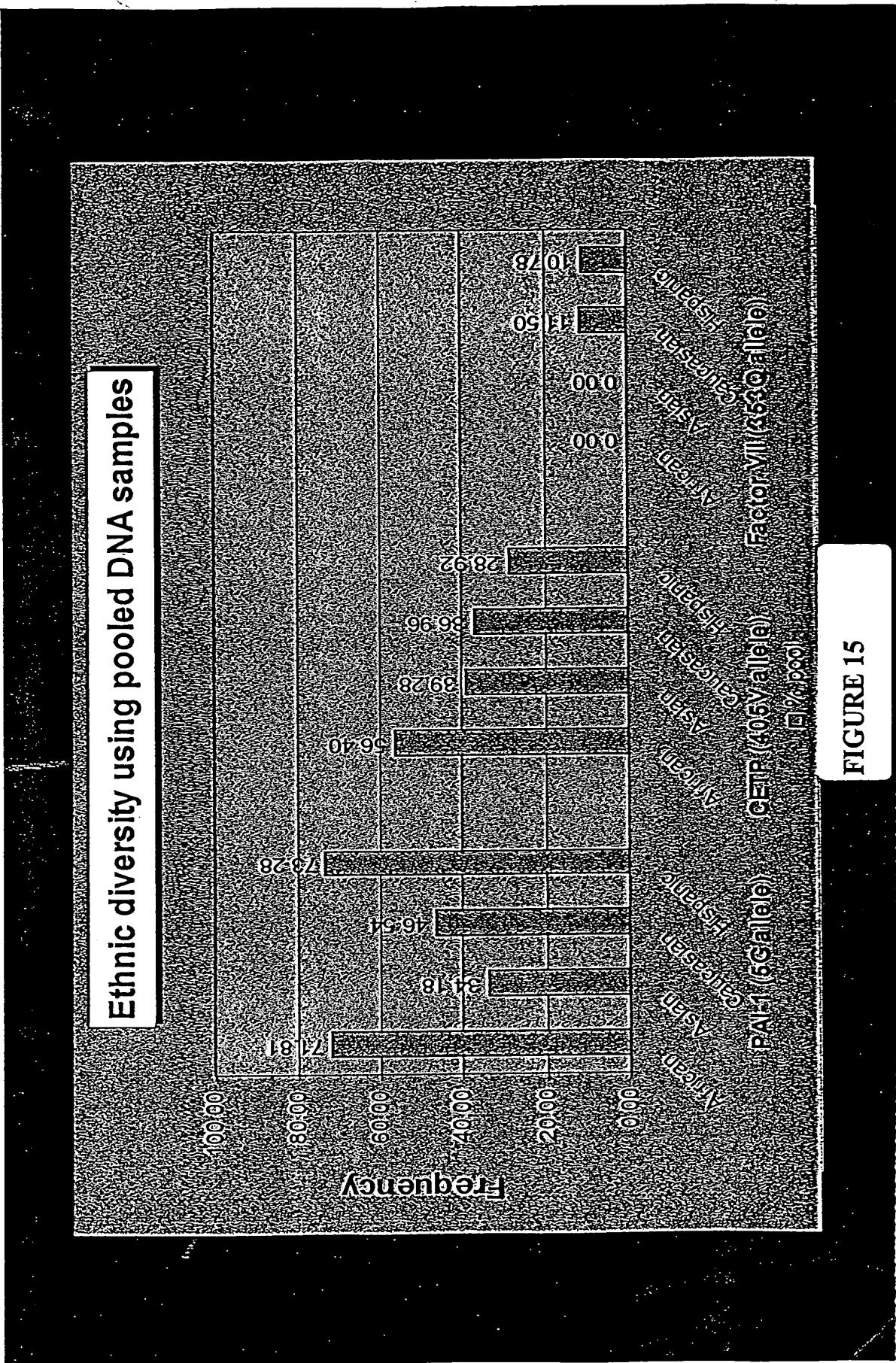
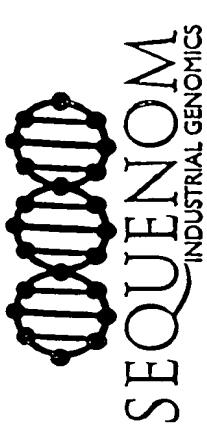


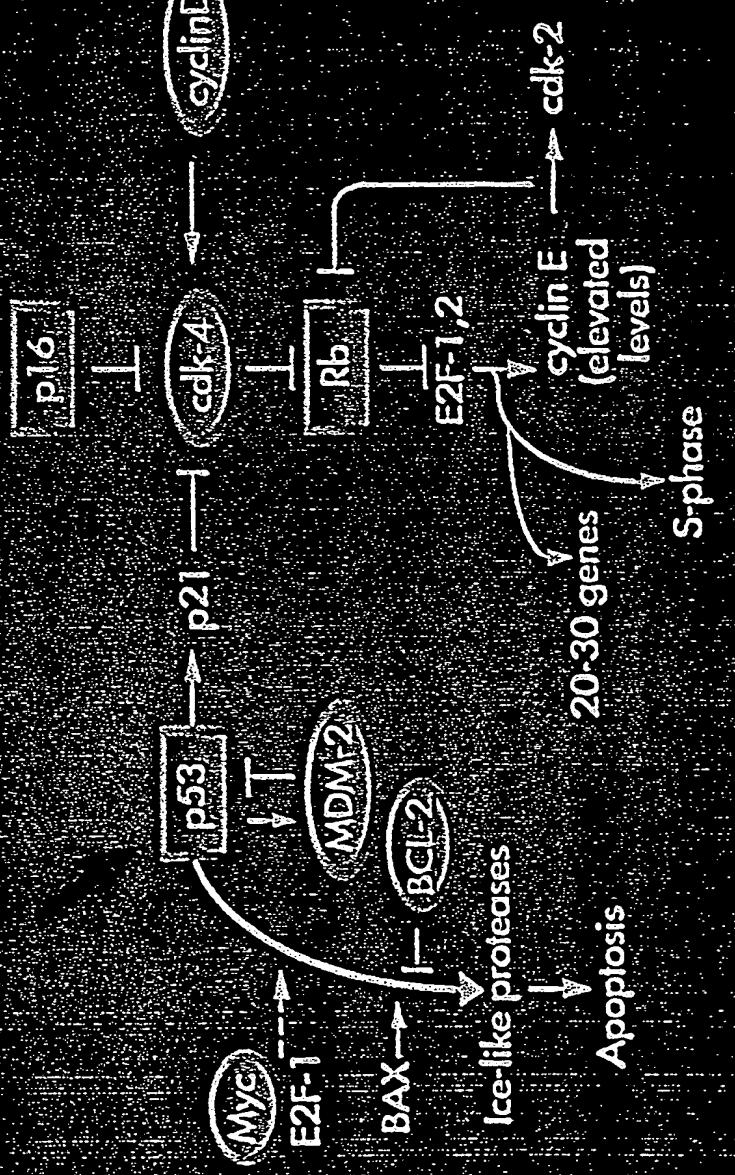
FIGURE 14

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# DNA MassArray™ Ethnic Diversity



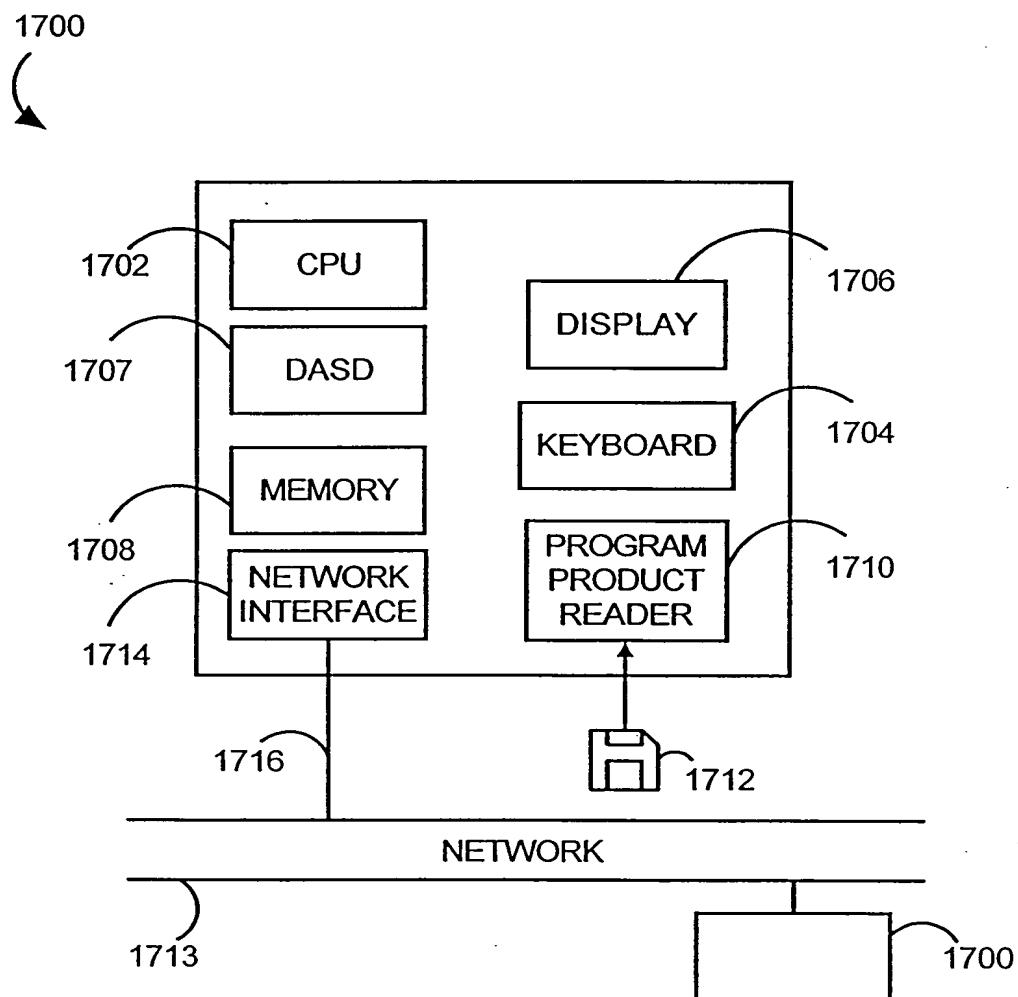
### P53-Rb Pathway



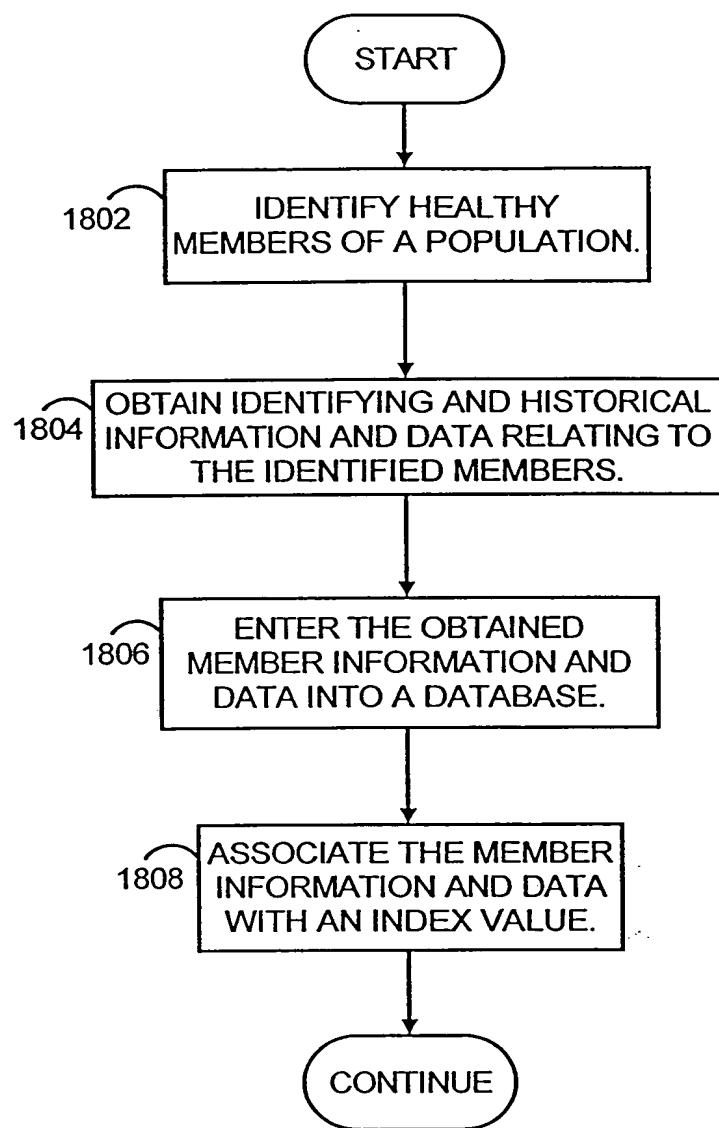
Levin A. Cell, 88: 323-331, 1997

FIGURE 16

00037000 2014-09-06



**FIGURE 17**



**FIGURE 18**

clone chromosome 17 (#48319)

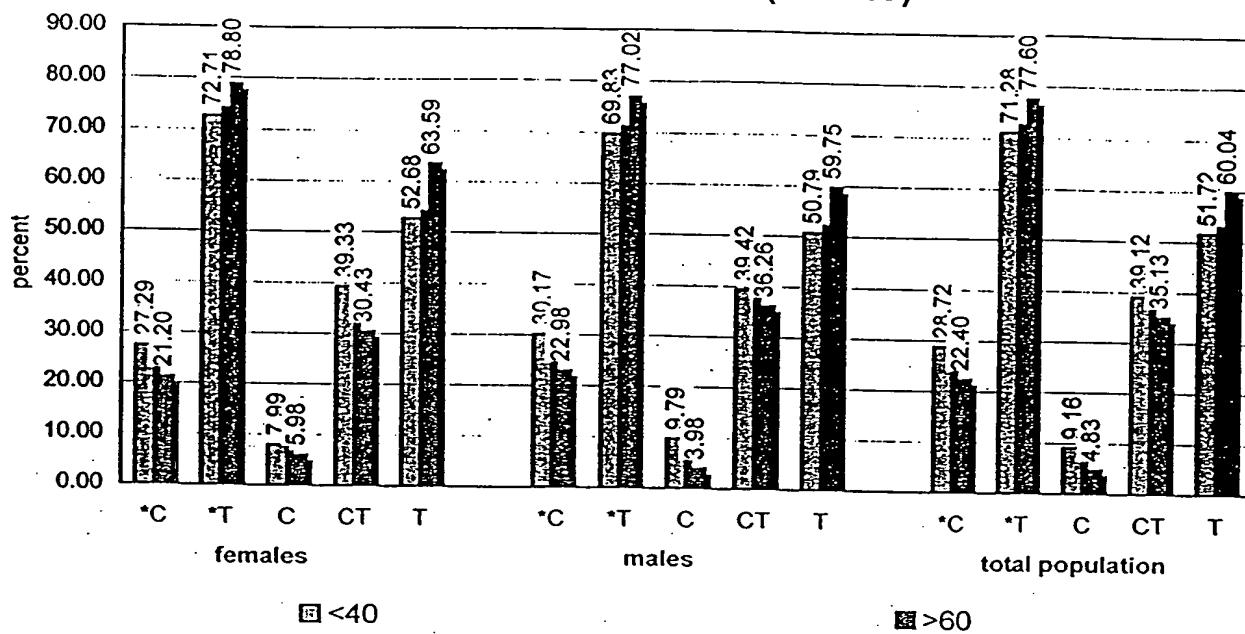


Figure 19

AKAP10 Ile646Val

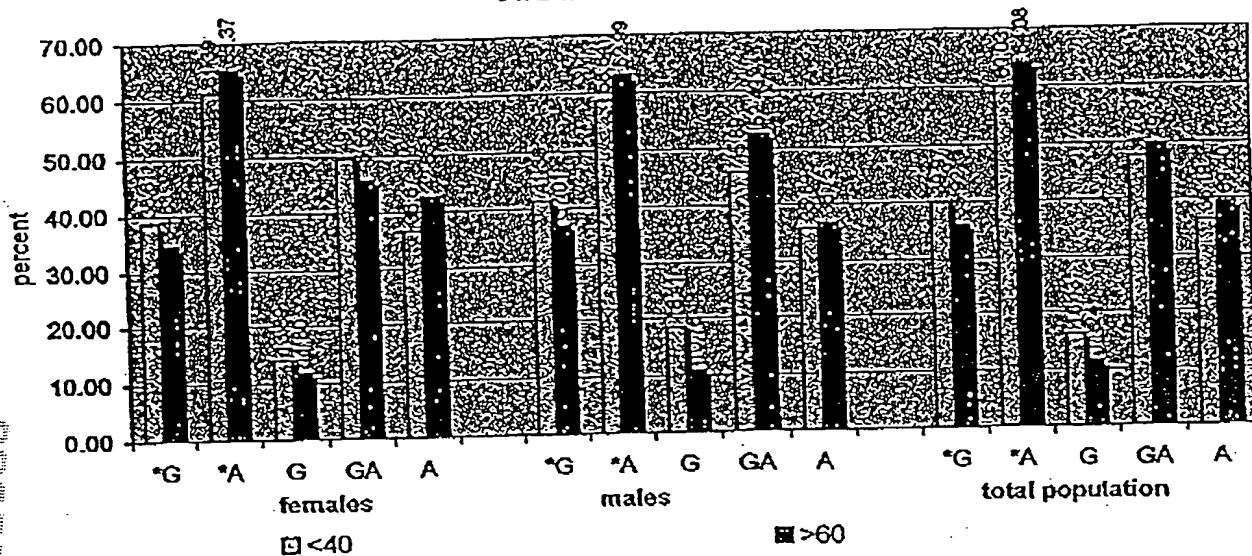
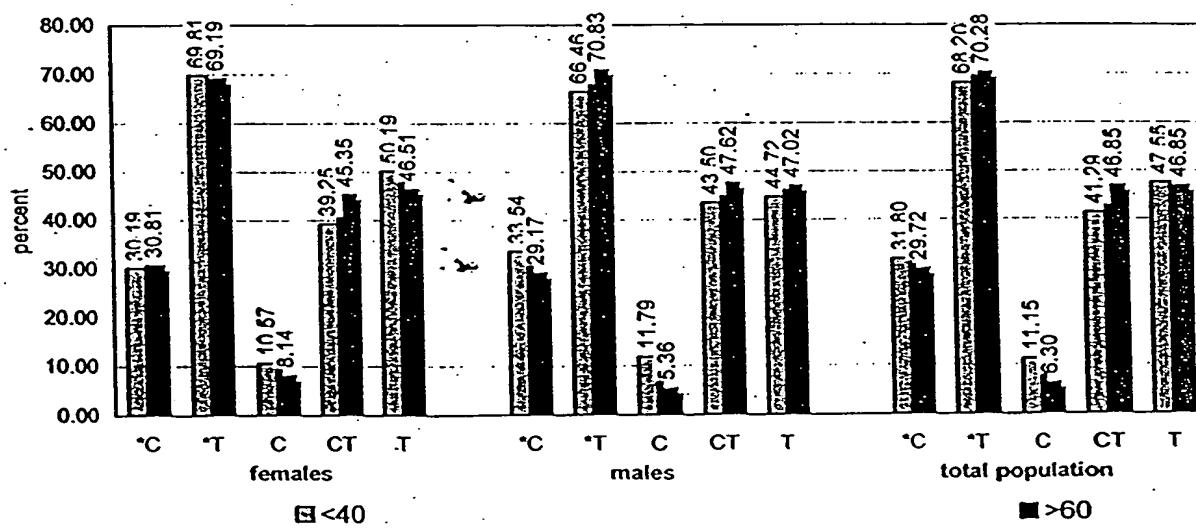


Figure 20

Figure 21

## methionine sulfoxid reductase A (#63306)



Bar Code

### **Donor Information**

Date of Birth		Height		Weight (lb)	
Month	Year	Ft	Inches		
JAN	1 9				
FEB					
MAR					
APR					
MAY					
JUN					
JUL					
AUG					
SEP					
OCT					
NOV					
DEC					

**Sex:**

Male       Female

<b>What physical activity do you do on a regular basis?</b>	<b>Are you a vegetarian?</b>
<input type="checkbox"/> Running <input type="checkbox"/> Swimming <input type="checkbox"/> Biking <input type="checkbox"/> Gymnastics <input type="checkbox"/> Other <input type="checkbox"/> None	<input type="checkbox"/> Yes <input type="checkbox"/> No

**Female:**

**How many times have**

**How many times have you been pregnant?      How many times did you give birth?**

To the best of your knowledge, what is the Ethnic Origin of your:

**Father    Mother**

- Caucasian (please mark specific geographic area below if known)  
 Northern Europe (Austria, Denmark, Finland, France, Germany, Netherlands, Norway, Sweden, Switzerland, UK)  
 Southern Europe (Greece, Italy, Spain, Turkey)  
 Eastern Europe (Czechoslovakia, Hungary, Poland, Russia, Yugoslavia)  
 Middle Eastern (Israel, Egypt, Iran, Iraq, Jordan, Syria, Other Arab States)

## African-American

- Hispanic (please mark specific geographic area below if known)**

Mexico

Central America, South America

Cuba, Puerto Rico, other Caribbean

**Asian (please mark specific geographic area below if known)**

- Asian (please mark one)  
 Japanese  
 Chinese  
 Korean  
 Vietnamese  
 Filipino

## **Native American**

Other  
Don't know

**How long  
have you  
lived  
there?**

Years	
0	0
1	1
2	2
3	3
4	4
5	5
6	6
7	7
8	8
9	9

**What is your highest grade you completed in school?**

- less than 8th grade  
 8th, 9th, 10th, or 11th grade  
 high school graduate or  
     equivalency  
 some college, 2 yr degree  
 college graduate, 4 yr degree  
 post graduate education or  
     degree

Mother Deceased? . Cause of Death Mother:

- |                              |       |               |
|------------------------------|-------|---------------|
| <input type="checkbox"/> Yes | ≤ 29  | Heart Disease |
| <input type="checkbox"/> No  | 30-39 | Cancer        |
|                              | 40-49 | Stroke        |
|                              | 50-59 | Accident      |
|                              | 60-69 | Suicide       |
|                              | 70-79 | Other, _____  |
|                              | 80-89 |               |
|                              | ≥ 90  |               |

**Father Deceased? Cause of Death Father**

- |                              |       |  |
|------------------------------|-------|--|
| <input type="checkbox"/> Yes | ≤ 29  | <input type="checkbox"/> Heart Disease |
| <input type="checkbox"/> No  | 30-39 | <input type="checkbox"/> Cancer        |
| If Yes at<br>what age?       | 40-49 | <input type="checkbox"/> Stroke        |
|                              | 50-59 | <input type="checkbox"/> Accident      |
|                              | 60-69 | <input type="checkbox"/> Suicide       |
|                              | 70-79 | <input type="checkbox"/> Other, _____  |
|                              | 80-89 |  |
|                              | ≥ 90  |  |

## FIGURE 22A







00000000000000000000000000000000

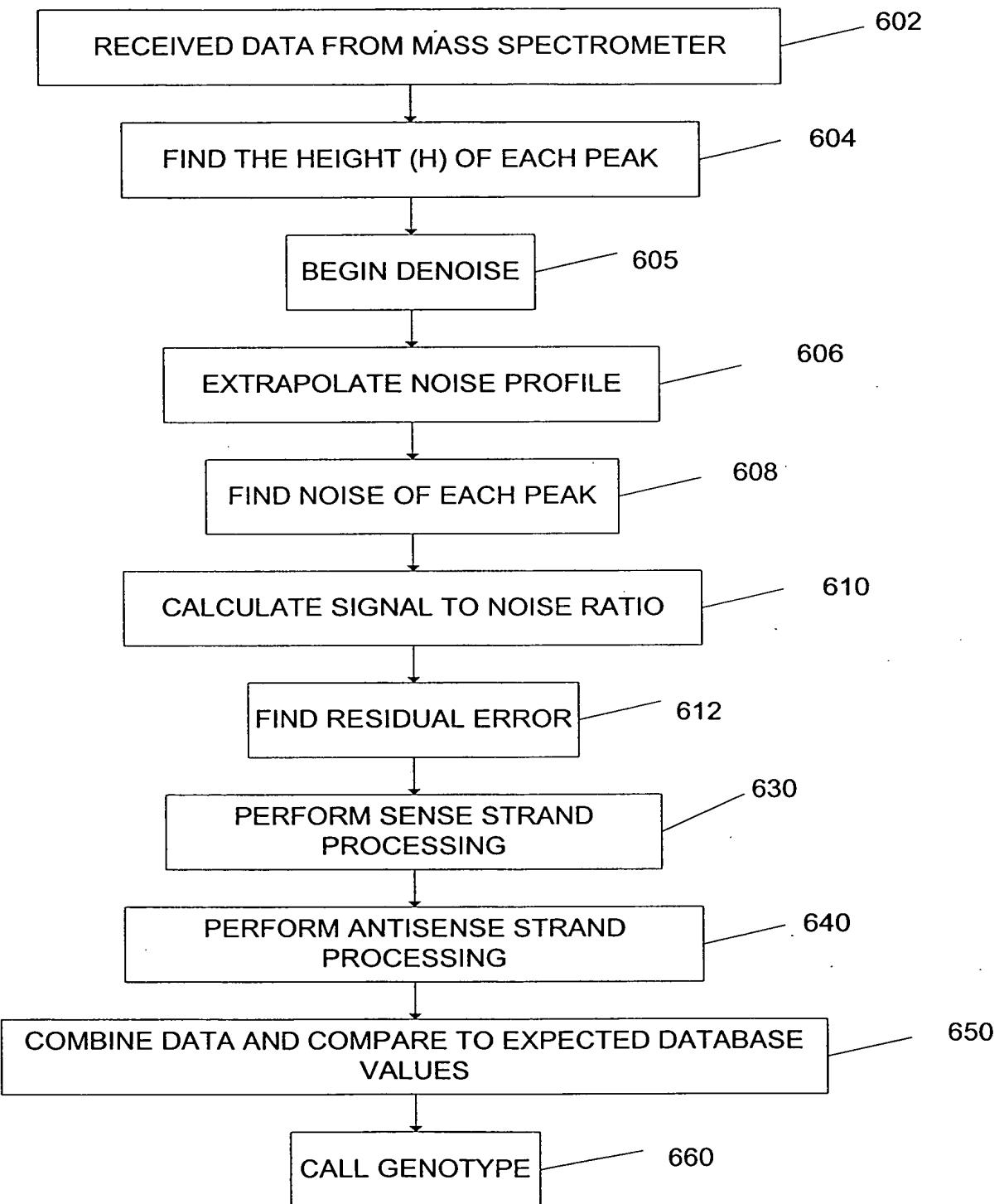


FIGURE 23

DO NOT "EFILE" 8/6/0

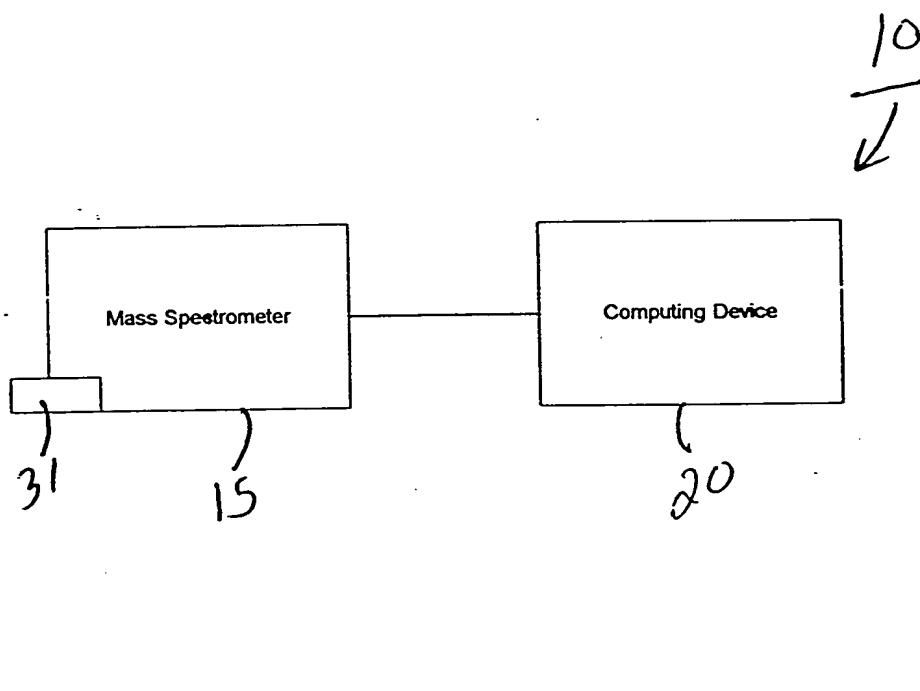
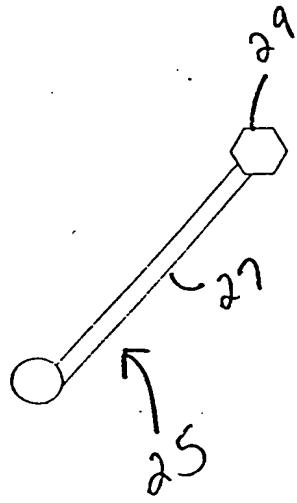
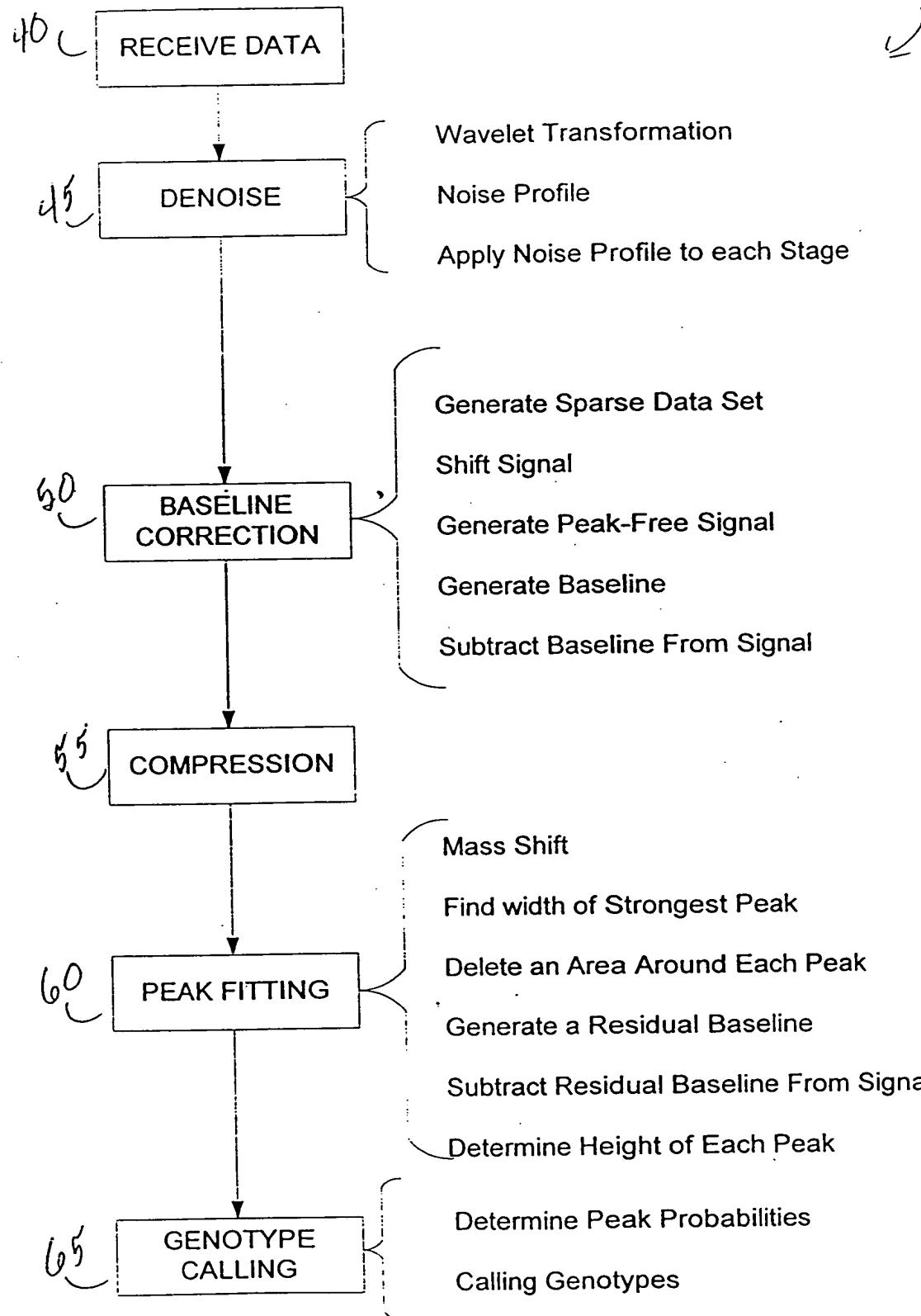
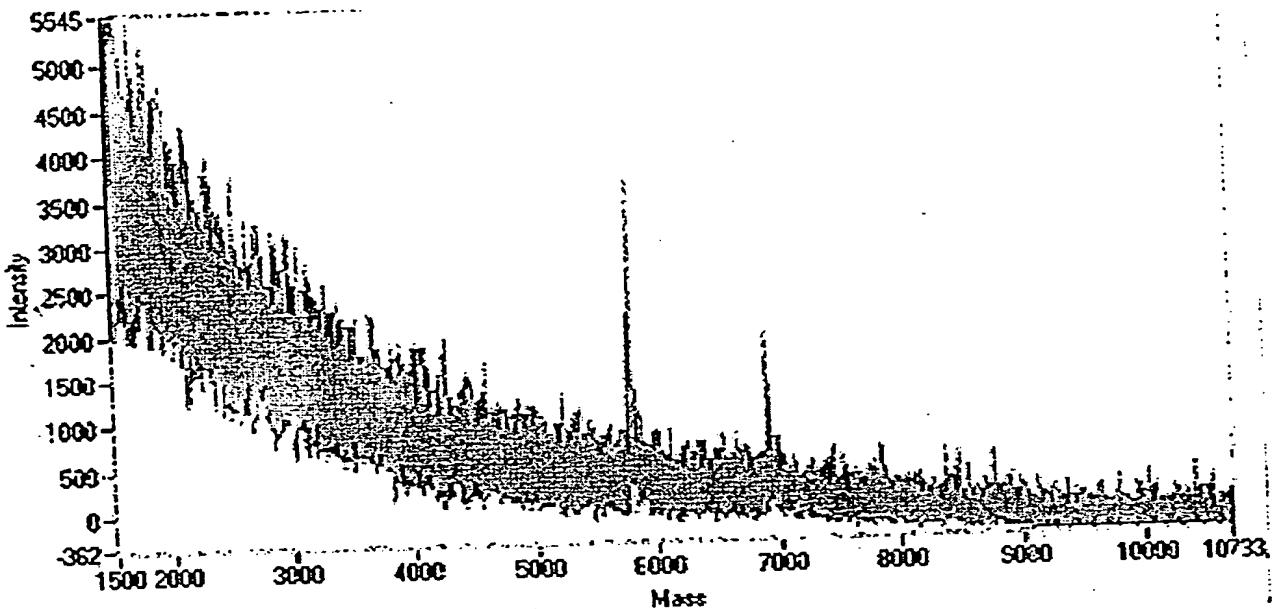


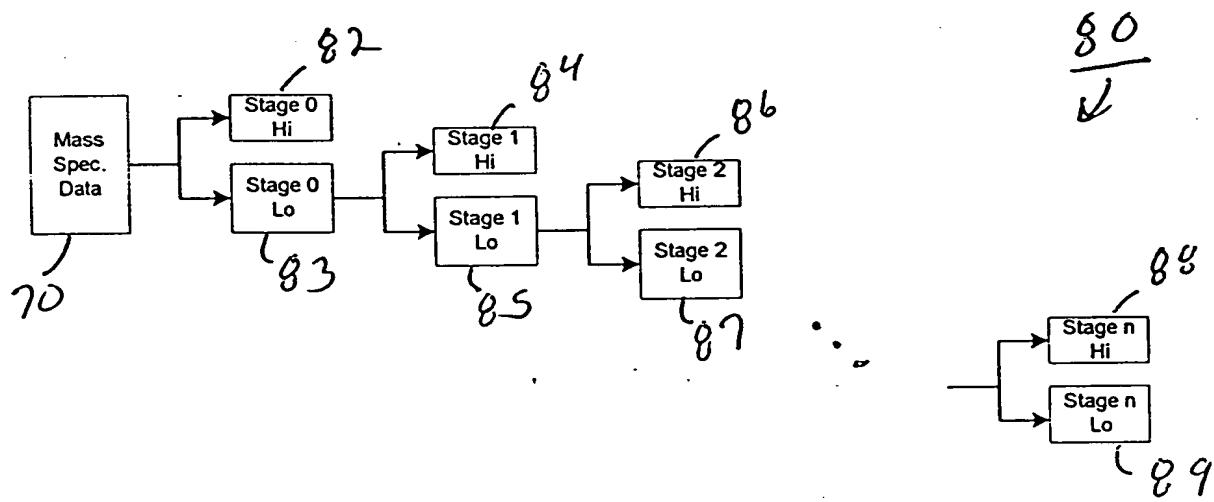
FIGURE 24

**FIGURE 25**

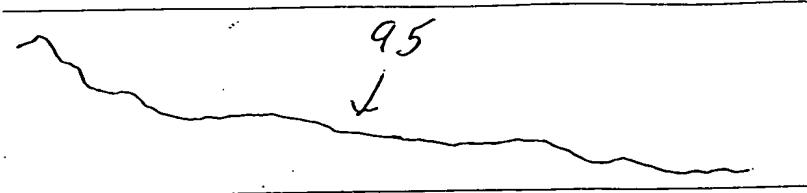


**FIGURE 26**

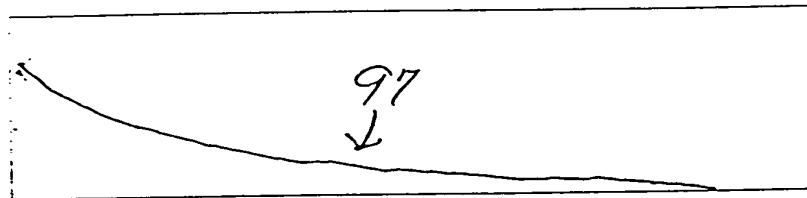
DRAFT DATE: 12/12/2010



**FIGURE 27**



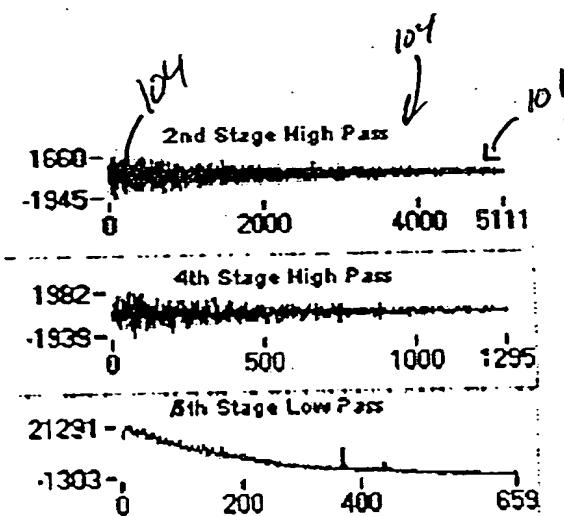
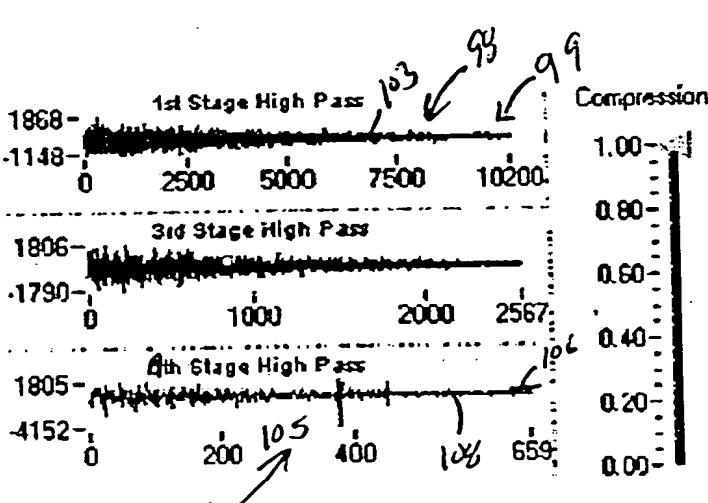
**FIGURE 28**



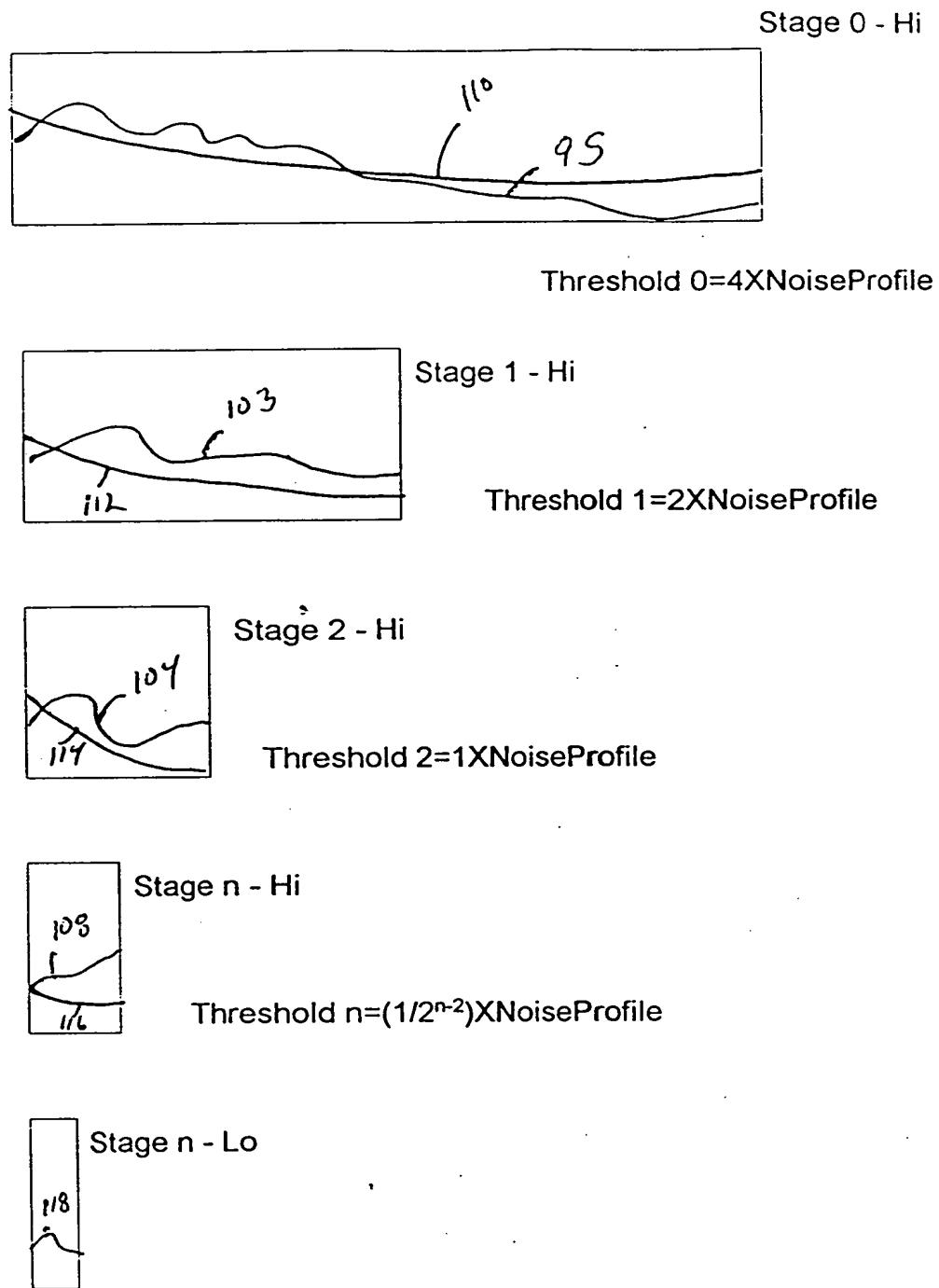
Exp fitting  
 $a_0 + a_1 \exp(a_2 m)$

**FIGURE 29**

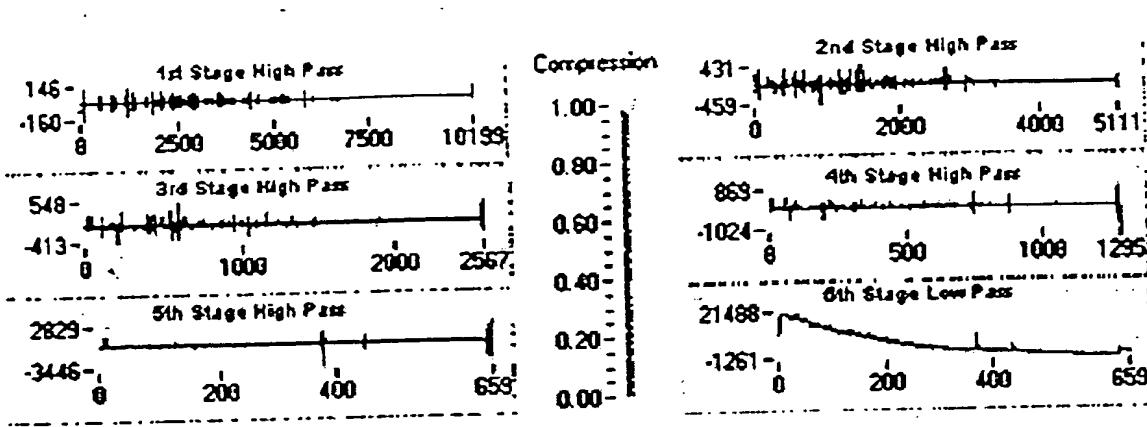
CODE FOR "E842C960"



**FIGURE 30**



**FIGURE 31**

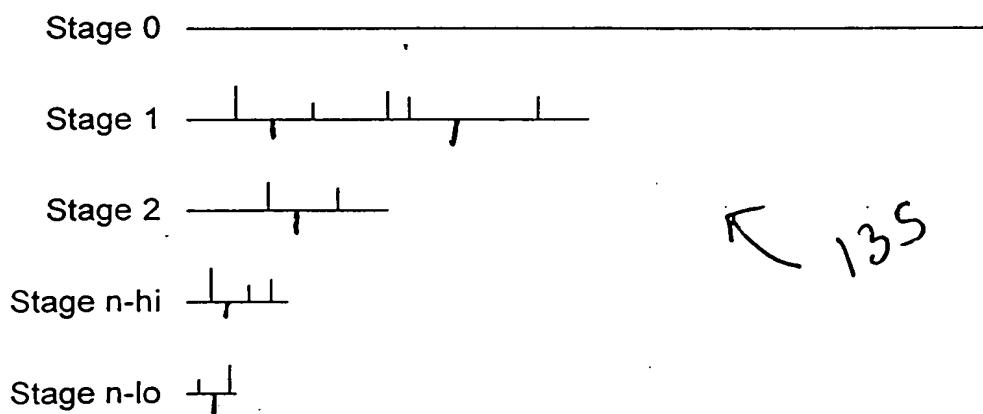


**FIGURE 32**

$$\text{Signal } (t) = \frac{(\text{Start } 0(t) + \text{Start } 1(t) + \text{Start } 2(t) \dots + \text{Start } 23(t))}{24}$$

**FIGURE 33**

SHIFT SIGNAL TO ACCOUNT FOR  
VARIATIONS DUE TO STARTING POINT



**FIGURE 34**

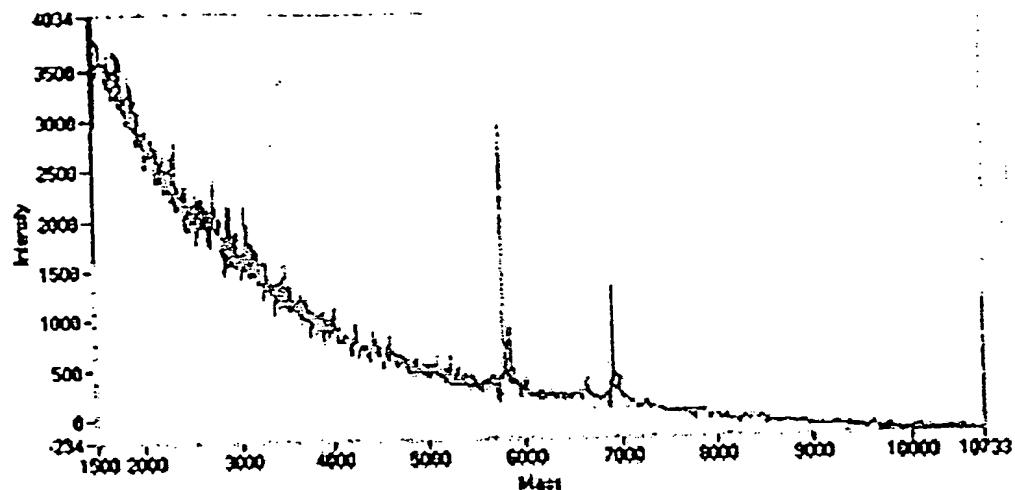


FIGURE 35

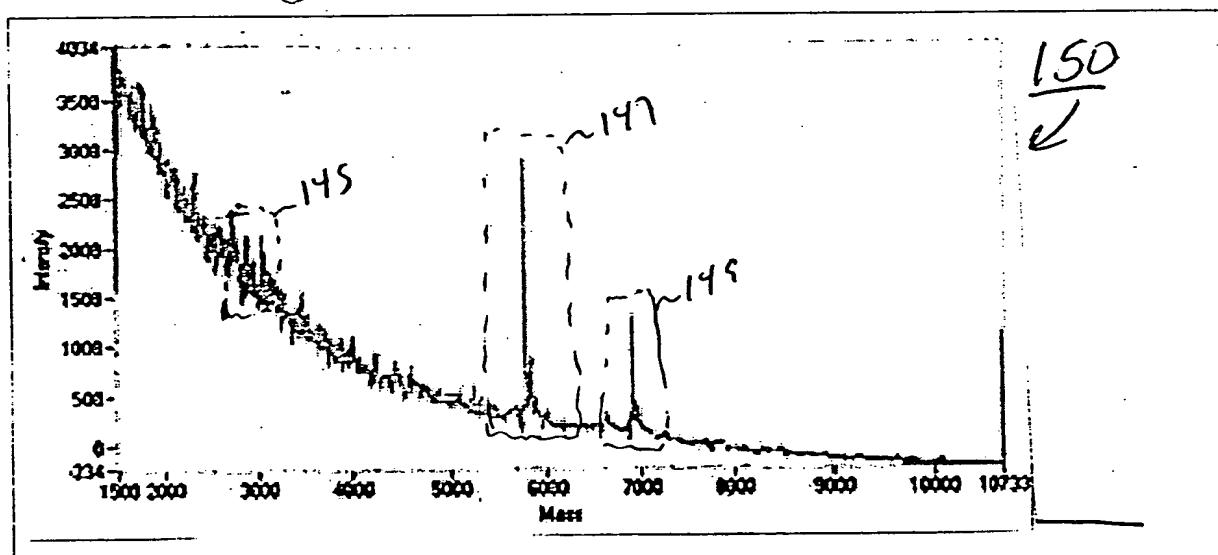
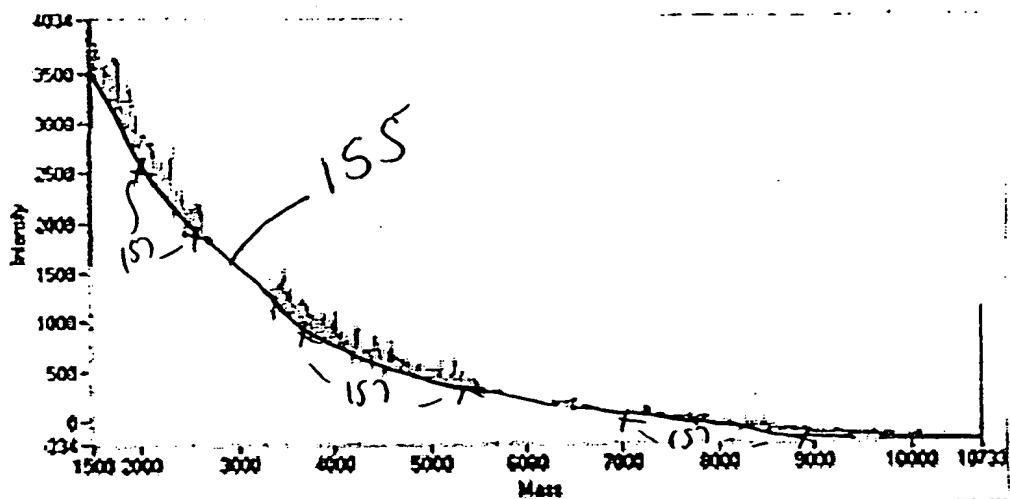
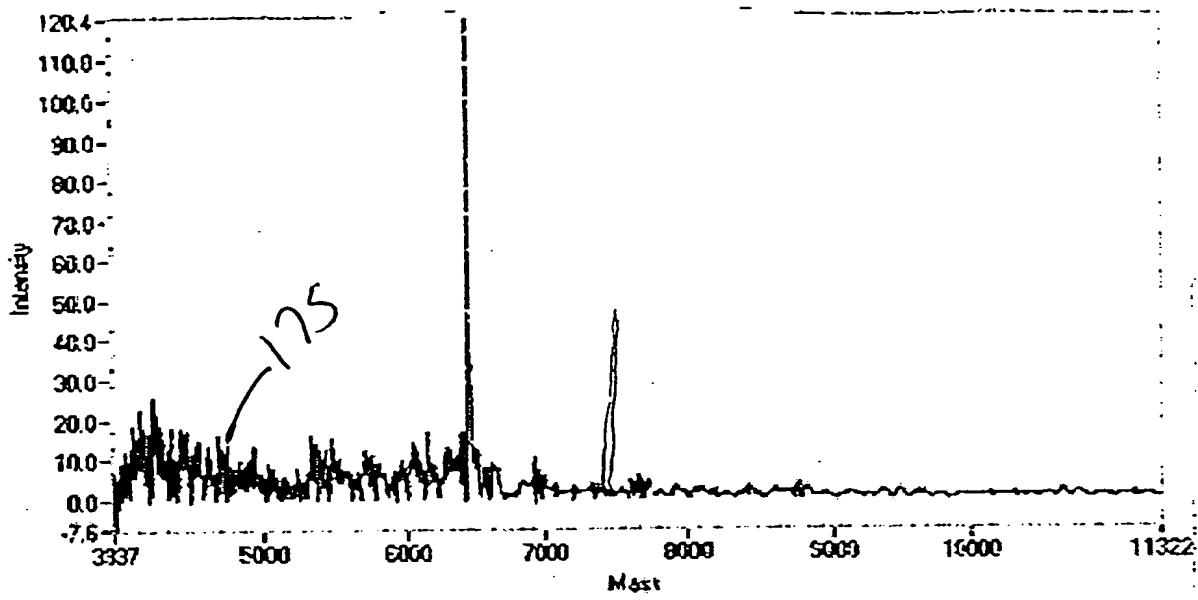


FIG. 13 - TAKE A MOVING AVERAGE, REMOVE SECTIONS EXCEDING A THRESHOLD

FIGURE 36



**FIGURE 37**  
FIND MINIMA IN REMAINING SIGNALS AND CONNECT TO FORM A PEAK FREE SIGNAL



**FIGURE 40**

NON-0 COEFFICIENTS	VALUE	B2	10^4	10^5	10^6	10^7	10^8	INTERMEDIATE	RELATIVE
100	25							100.025	100.025
150	220							150.220	50.220
500	.1							500.0001	350.0001
10,050	800							10,050.8	9550.8
10,075	890							10,075.89	25.89
11,125	910							11,125.91	150.91
12,100	1000 (MAX)							12,100.99999	975.99999
13,250	940							13,250.94	1150.94

## FIGURE 41

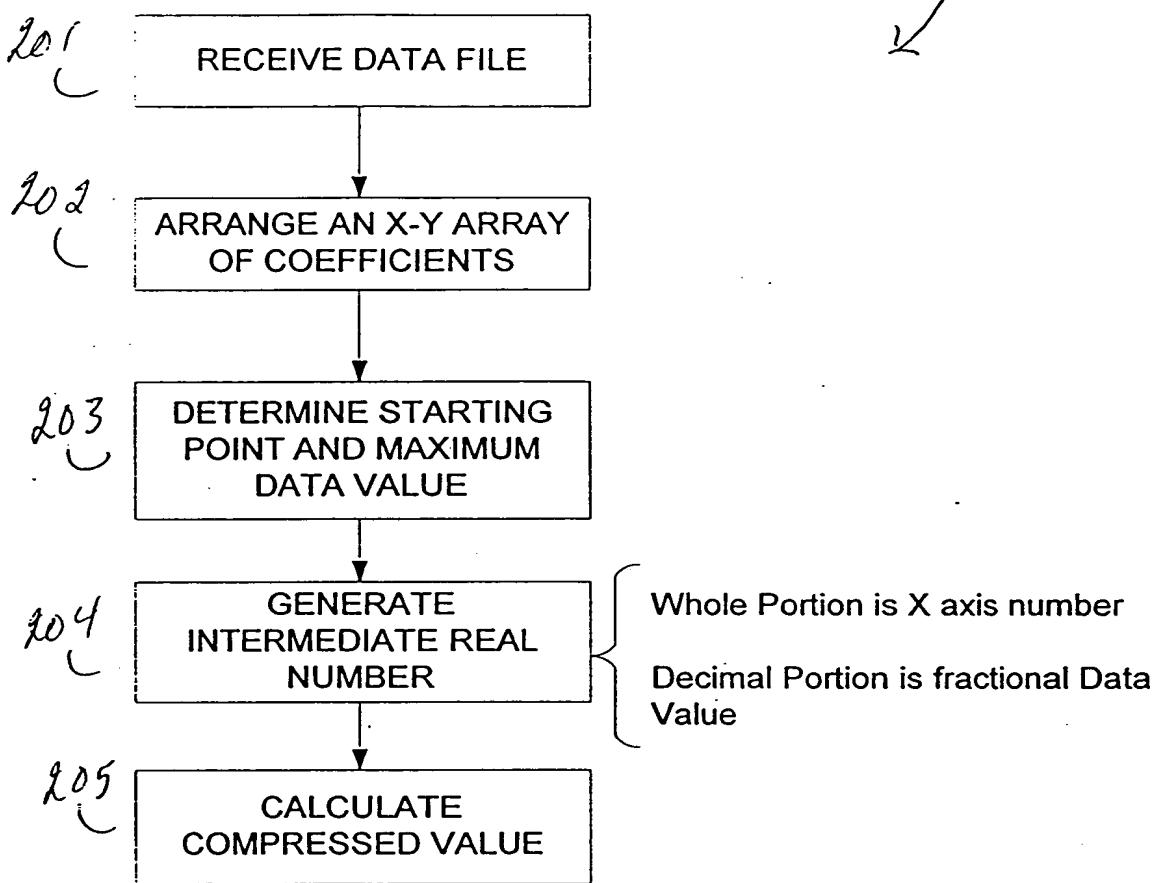


FIGURE 42

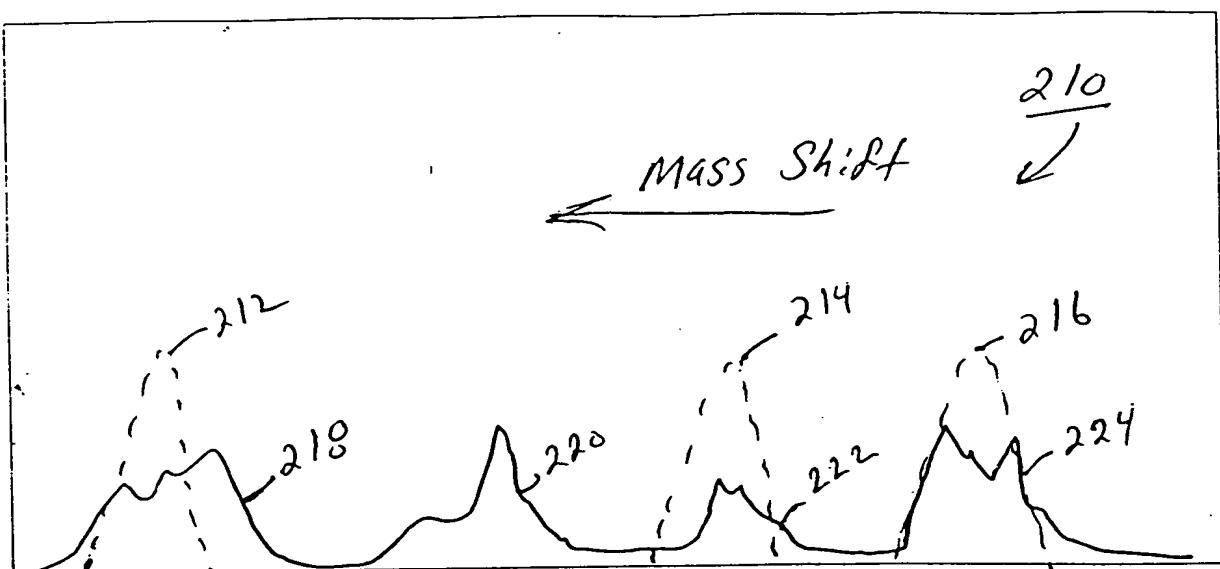


FIGURE 43

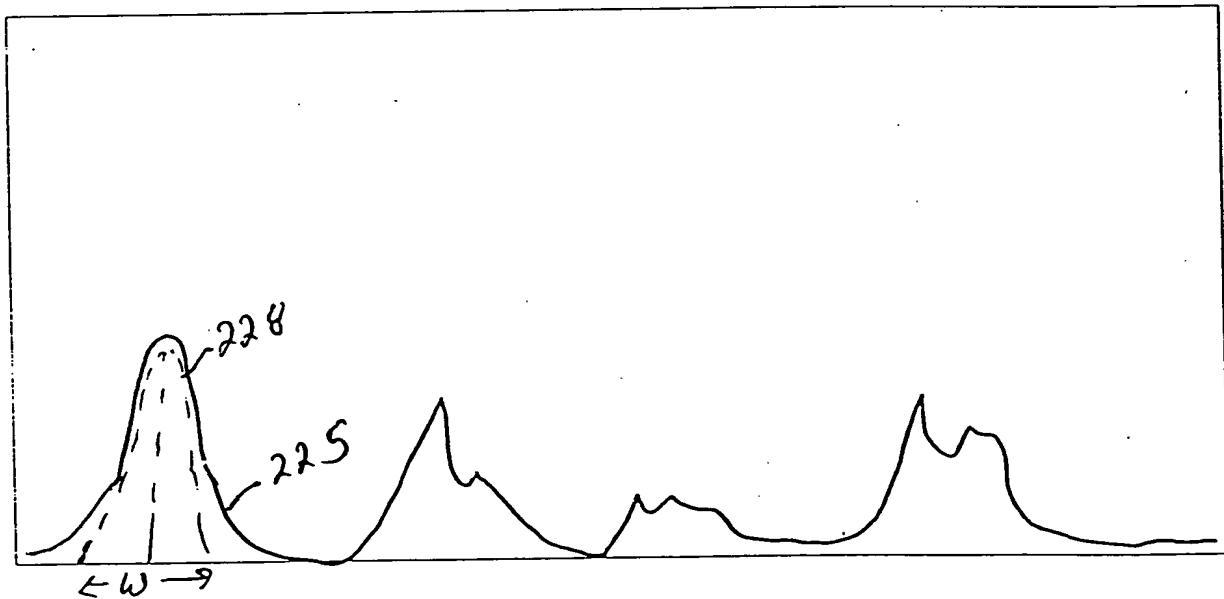


FIGURE 44

0968274633-101200

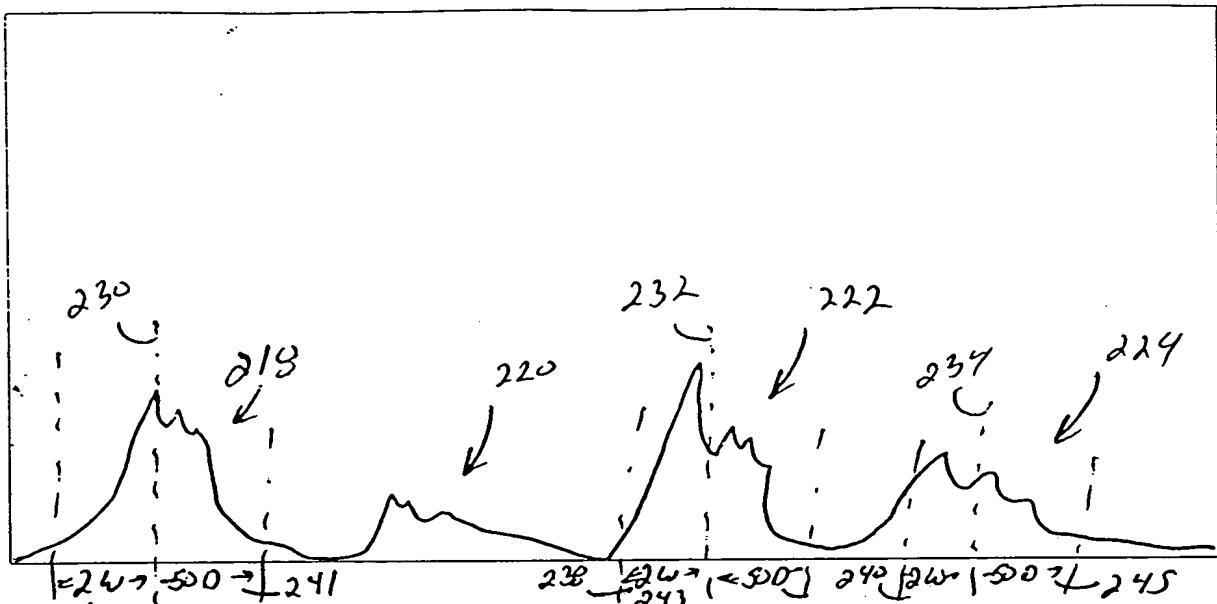


FIGURE 45

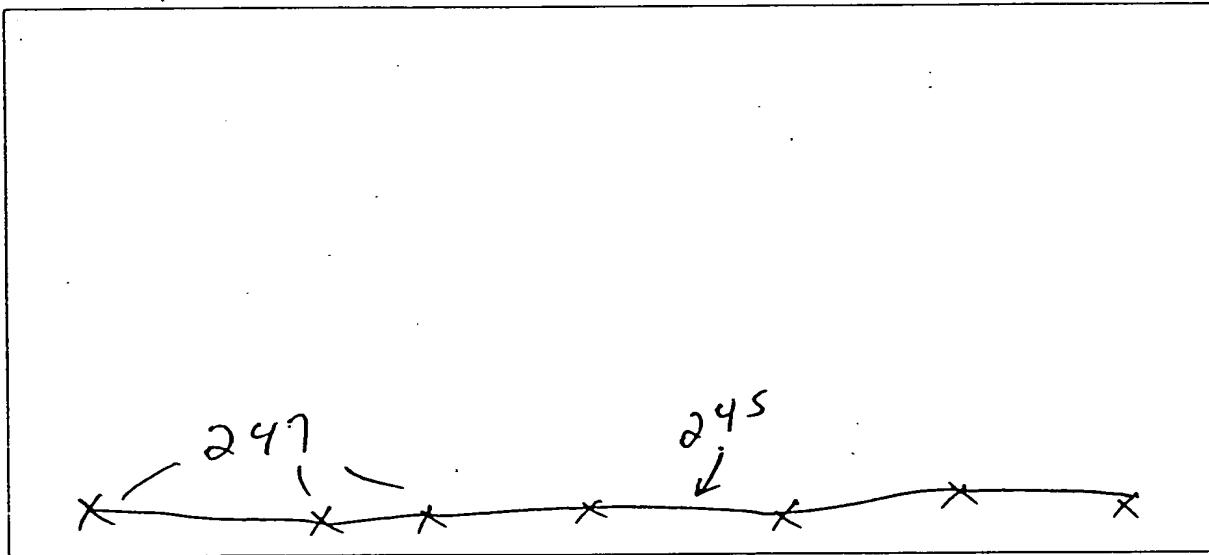


FIGURE 46

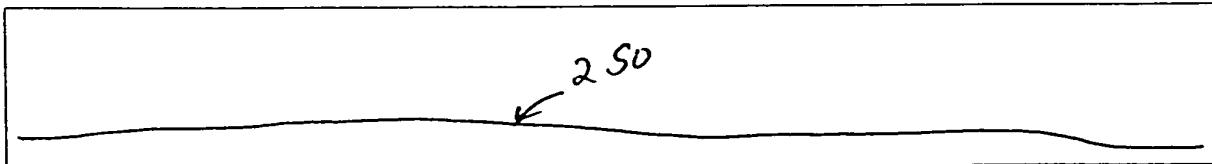
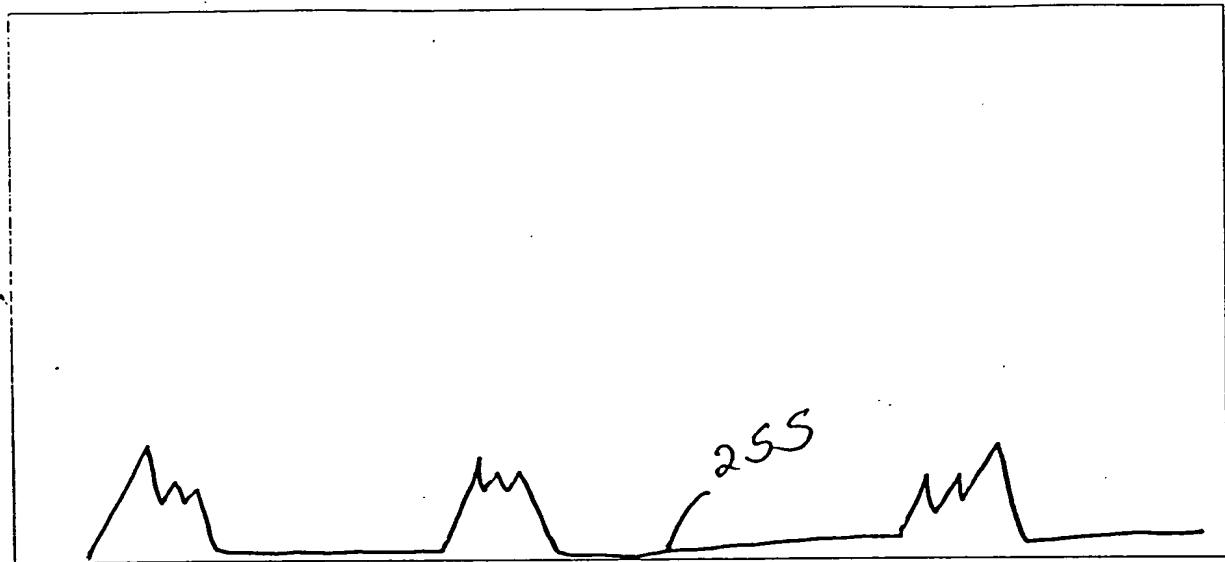
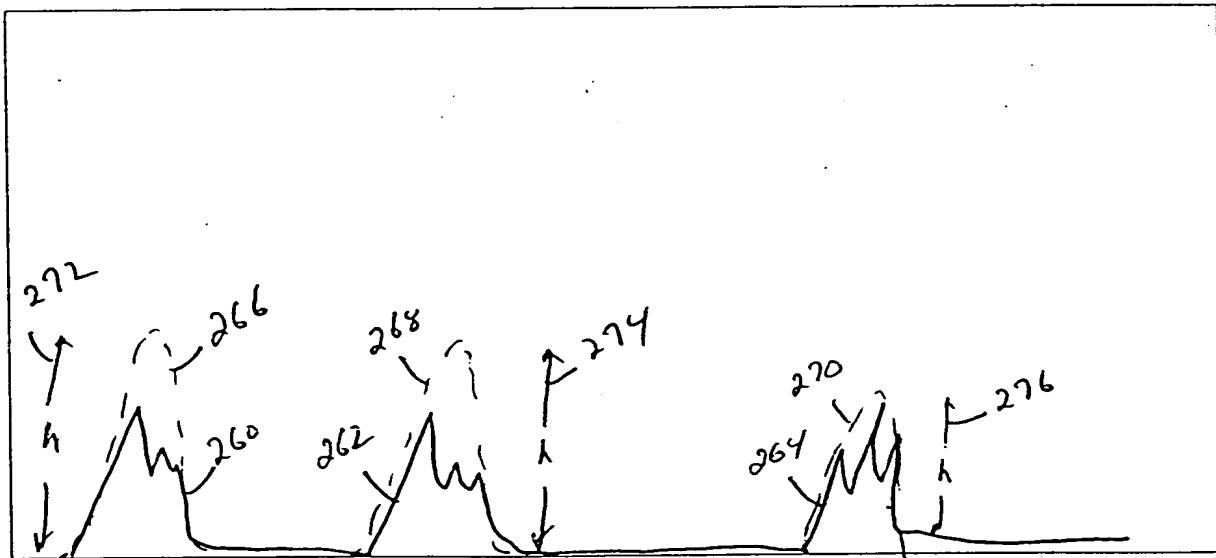


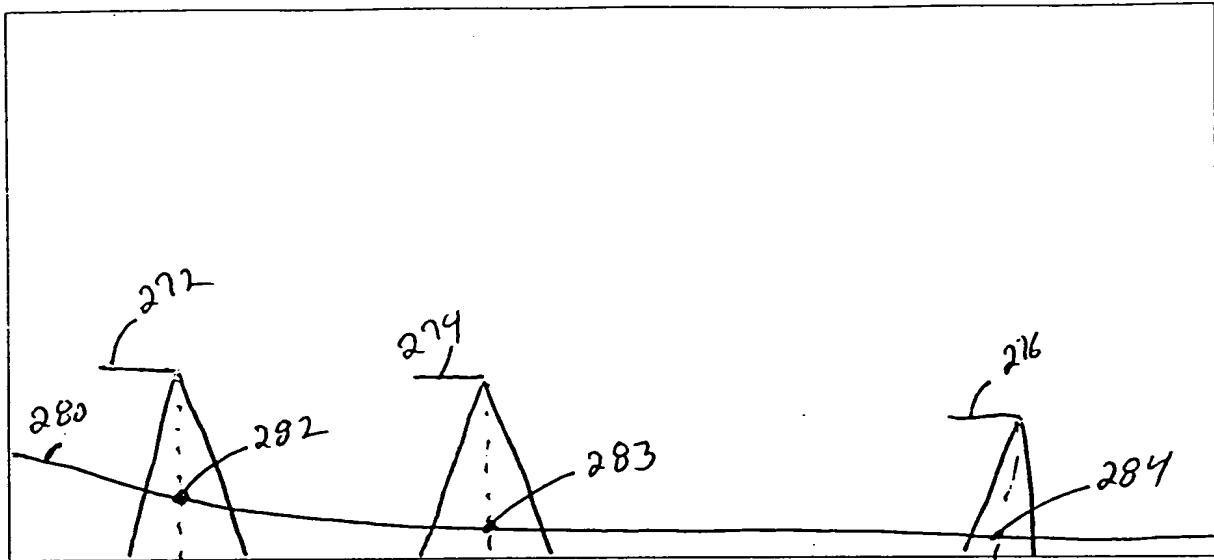
FIGURE 47



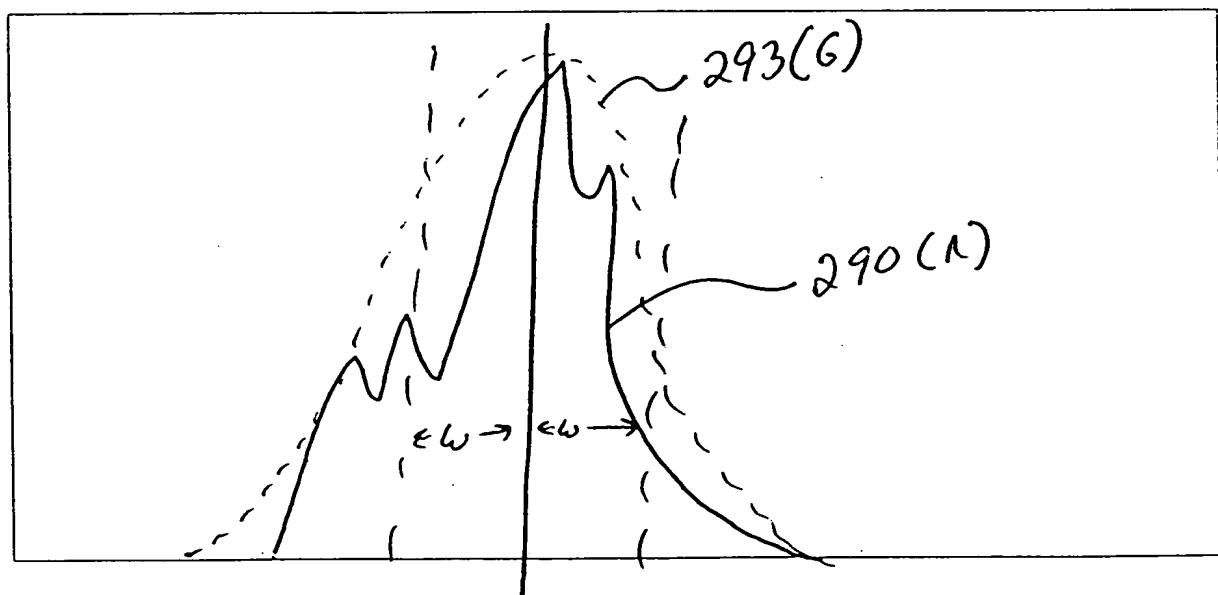
**FIGURE 48**



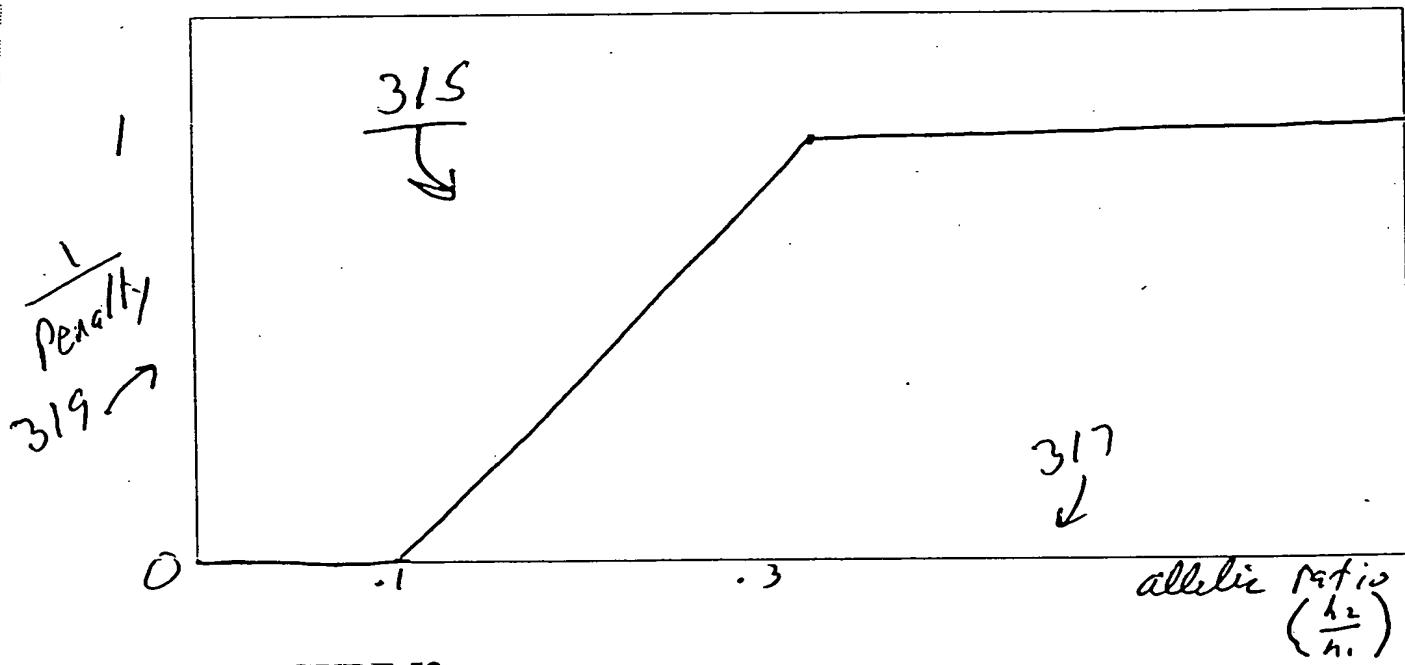
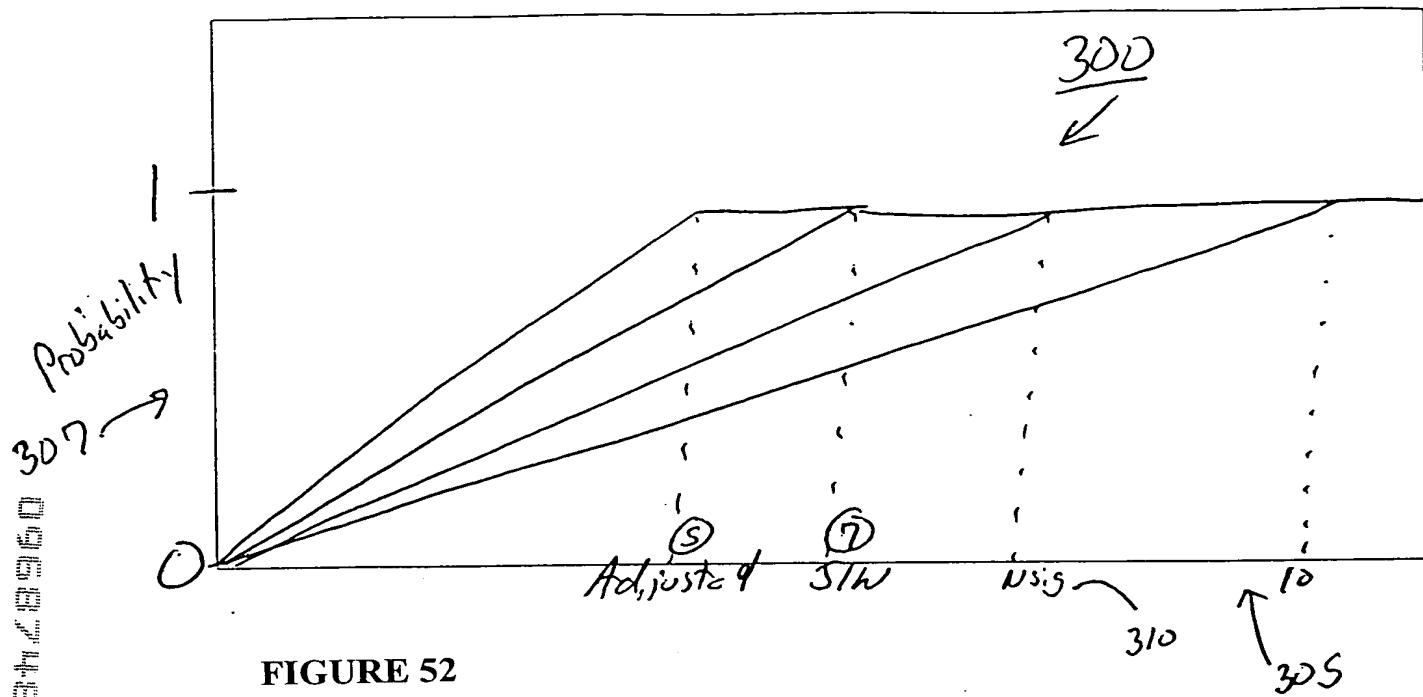
**FIGURE 49**

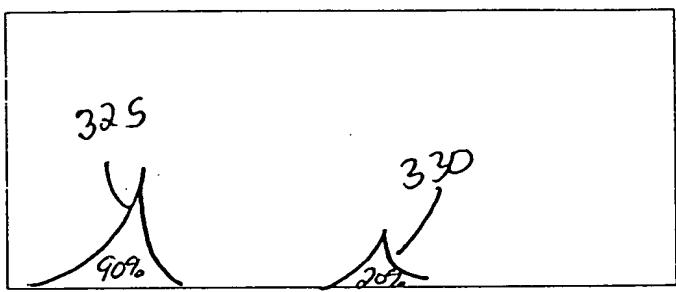


**FIGURE 50**



**FIGURE 51**





**FIGURE 54**

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PROBABILITY OF GG EXISTING:

$$\begin{aligned} P(GG) &= P(G) * P(1-C) \\ &= 90\% * (100\% - 20\%) \\ &= 90\% * 80\% \\ &= 72\% \end{aligned}$$

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PROBABILITY OF CC EXISTING:

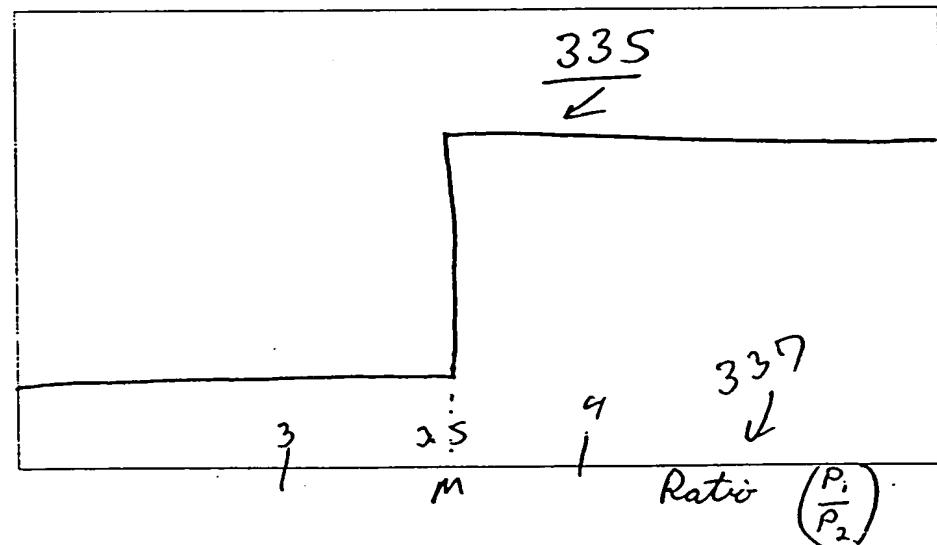
$$\begin{aligned} P(CC) &= P(C) * P(1-G) \\ &= 20\% * (100\% - 90\%) \\ &= 20\% * 10\% \\ &= 2\% \end{aligned}$$

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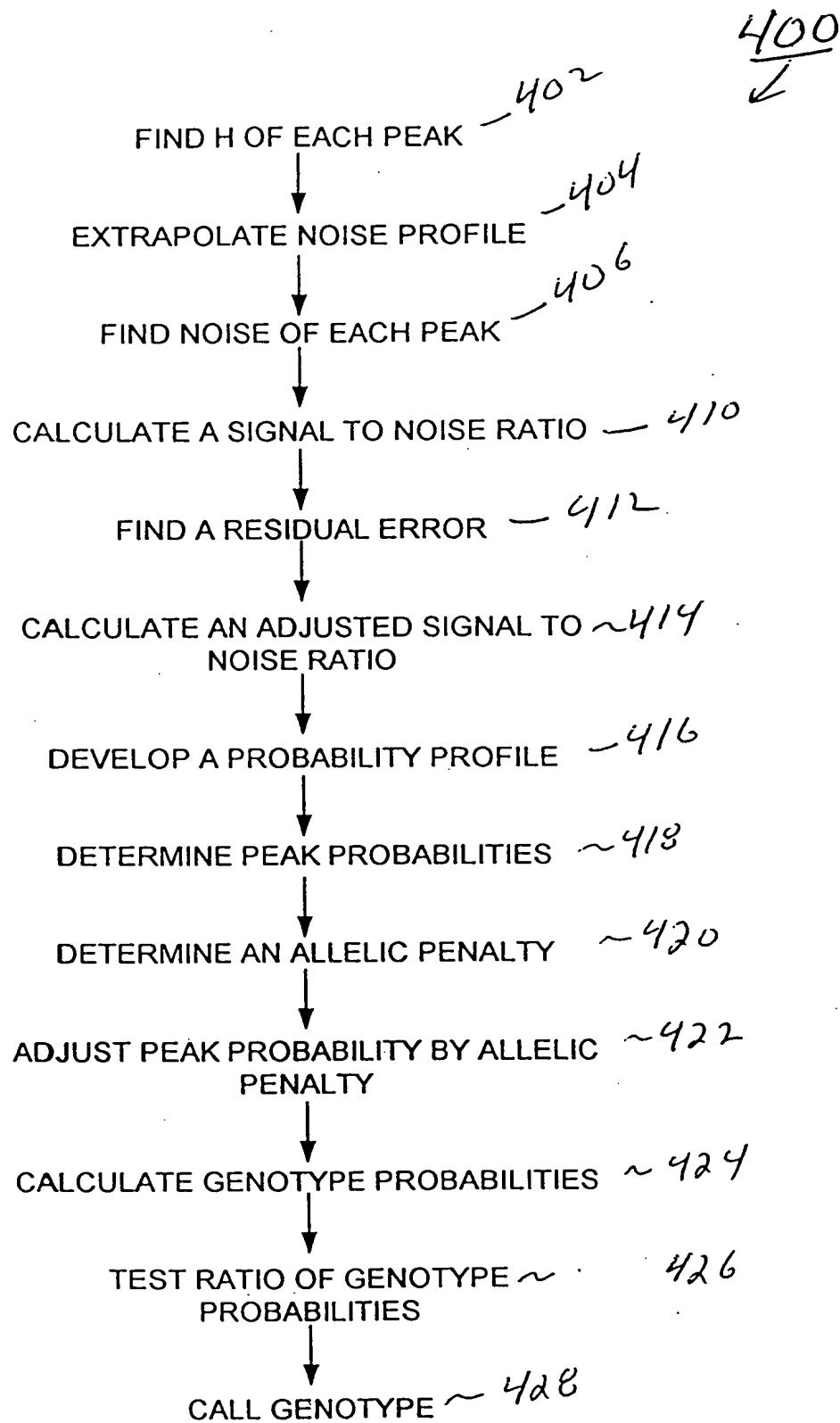
PROBABILITY OF GC EXISTING:

$$\begin{aligned} P(GC) &= P(G) * P(C) \\ &= 90\% * 20\% \\ &= 18\% \end{aligned}$$

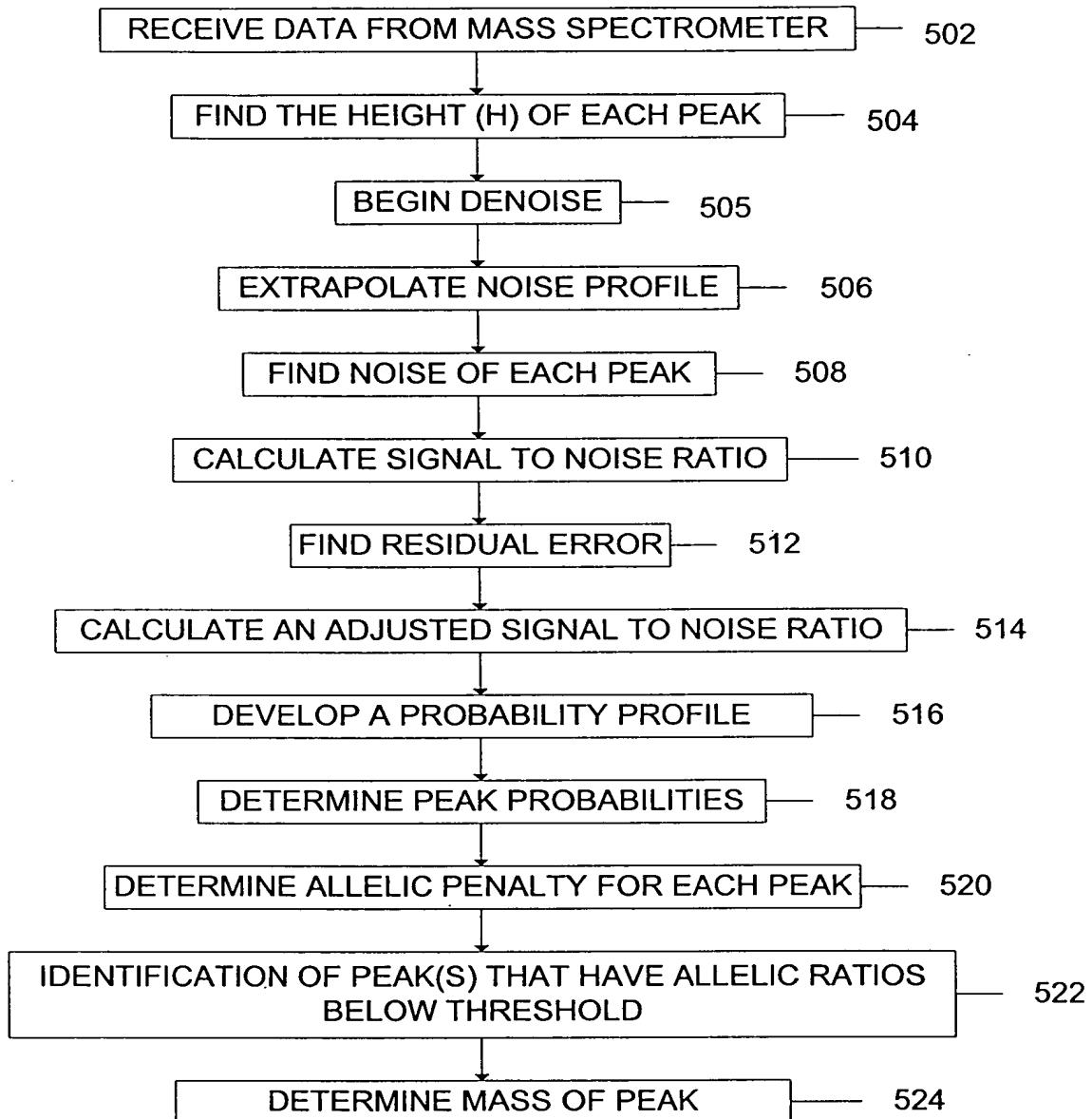
096621662-1011300



**FIGURE 55**

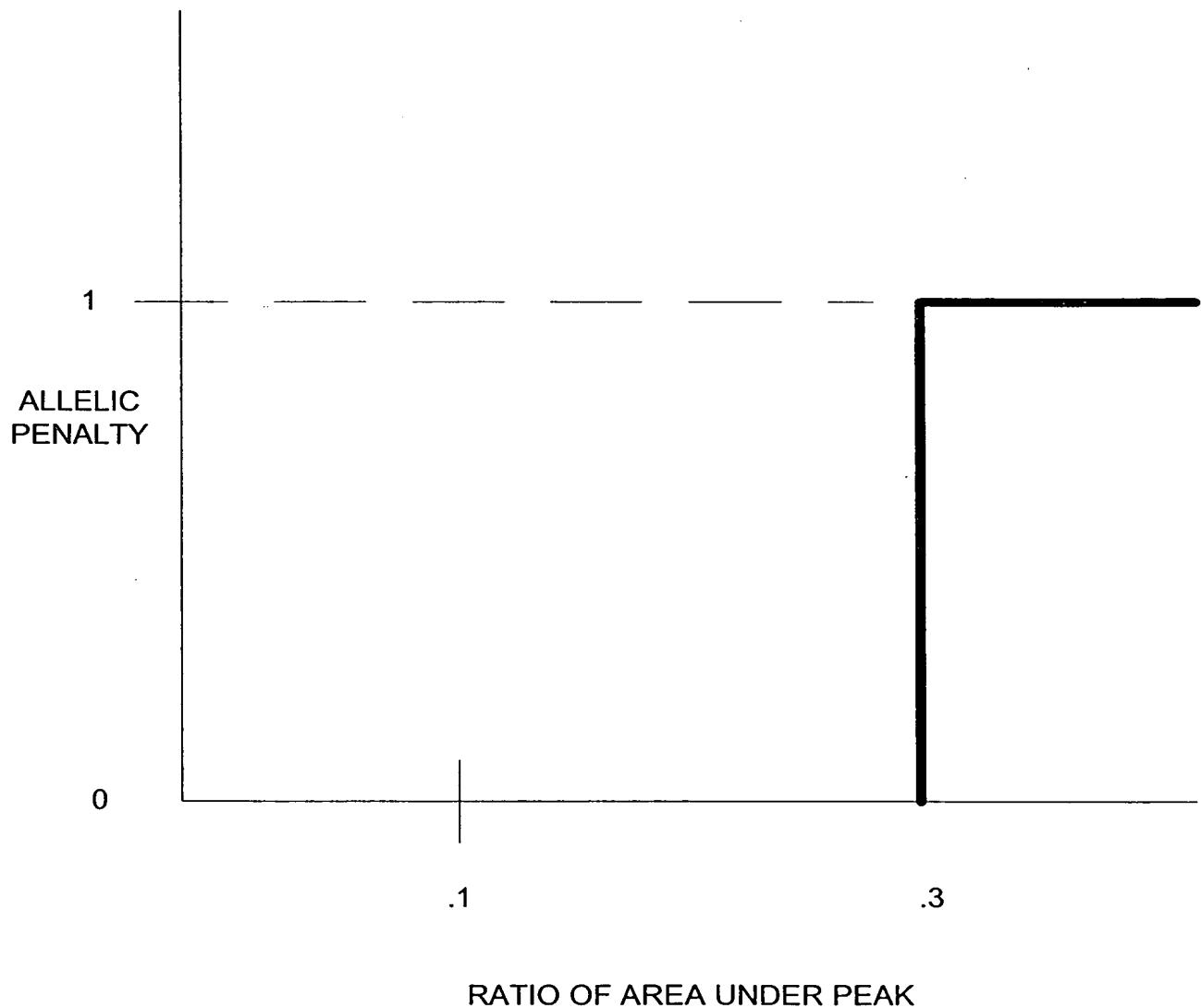


**FIGURE 56**



**FIGURE 57**

FIGURE 58  
1001300



**FIGURE 58**